CASEBOOK OF

Clinical Neuropsychology

Edited by

Joel E. Morgan
Ida Sue Baron
Joseph H. Ricker

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For J. E. M.: To Steffie and Freddie
For I. S. B.: To Peter, David, and Cara
For J. H. R.: To Lance

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Foreword

Most will agree that what has now evolved into the field of clinical neuropsychology was initially built on a foundation of single case studies. The unfortunate fates of Phineas Gage, Tan (Broca aphasia), and H. M. have captured imaginations of multiple generations of students entering the field. The value of these famous cases lies well beyond what might be considered answers to a modern game of "neuroscience trivia." Such cases actually serve to provide us with a template for developing a detailed understanding of the effects of brain dysfunction, not only on behavior, but on the person as a whole.

While neuropsychology as a science owes much of its success to findings from studies on patient groups, the individual case study continues to play a significant role in advancing our knowledge in both research and clinical training. Our science will continue to move along with continued descriptions of unusual cases that challenge our theories and lead to paradigm shifts. However, a well-prepared case presentation is equally valuable to our field because it has the potential to not only instruct young students in the use of neuropsychological methods but also to provide seasoned clinicians with a means to refine their understanding of patients seeking their services.

Cognitive scientists have told us for years that categorical learning proceeds through exposure to a series of specific exemplars. The model of clinical training used by neuropsychologists and other health professional fields follows this route as it is based on a system of providing its novices with exposure to individual cases by observation and through direct clinical contact until they reach a point where they are able to provide the service on their own. This forms the basis of the old adage "watch one, do one, and teach one." Much of our personal knowledge of clinical phenomenology is clearly anchored by the memorable cases we see.

This book provides an insightful look into a number of clinical syndromes through the case presentation method. The editors have assembled an all-star cast of neuropsychologists with expertise in a wide range of clinical subspecialties. While the volume includes descriptions of some relatively rare syndromes, it also includes many excellent examples of the experts' approaches to what might be considered by many to be rather routine cases. The result is an entertaining mix of chapters that provides the reader with the important insights on what is typical in neuropsychology in addition to instruction on how to approach those types of cases seen less frequently. The reader will enjoy these well-written accounts of real-life applications of clinical practice, which provide an inside view of the richness of the data obtained through a comprehensive and evidence-based approach to neuropsychological assessment.

William B. Barr, PhD, ABPP
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Preface

Clinical evaluation is a basic function of the clinical neuropsychologist, as well as a demonstration of the art of clinical neuropsychology. While there is no substitute for the role of experience, even a novice practitioner may be expected to make astute observations and revealing insights in the service of clinical case formulation. To the dedicated clinician, the hands-on approach of clinical examination has no equal. While scientific revelations concerning brain–behavior relationships often emerge from well-controlled studies of large numbers of subjects, it is the challenge of the singular case that has an extraordinary attraction.

No doubt each clinical neuropsychologist has a profound professional memory related to at least one patient who had a particular disorder, whether it was a condition to be documented or detected. These memorable cases form what is arguably essential knowledge about the profession of clinical neuropsychology, and they underscore the core responsibilities that are basic to examination in the service of the individual.

How many times has a clinician been about to evaluate a patient but wished to preliminarily consult a knowledgeable colleague about what is known about the typical presentation, the most efficacious way to assess the patient, the kinds of recommendations/interventions that have proved useful…or, the solution to the diagnostic befuddlement engendered by the rare, singular case?

Recognizing the importance of these in vivo experiences, we decided to enlist the expertise of our colleagues and produce a casebook. The contributors were encouraged to pull a chart out of their files, one they could not forget or might favor for teaching. The range is from the ordinary to the unique. Authors were asked to detail the key facts related to the diagnosis at issue within a broadly set but logically organized framework and to provide their clinical data and interpretive formulations, with the aim that their case discussion have generalizable utility to colleagues.

Originally conceived as a companion to the *Textbook of Clinical Neuropsychology* (Morgan & Ricker, 2008, New York: Taylor & Francis), it is our intention that the *Casebook of Clinical Neuropsychology* serve as a reference textbook on which the reader may rely in preparation for examination of a patient with a known diagnosis, but whose disorder might not have been previously encountered by this clinician with any frequency. The volume may also prove helpful to compare and contrast one's own clinical findings and observations with those of a colleague who has had experience with the condition of interest. One can learn a good deal about diagnostic considerations and potential intervention strategies when these are described by informed and experienced clinical professionals. We certainly gained these insights as we read each of the contributed chapters.

The chapters are broadly grouped by general diagnostic categories. Child and adult cases are combined within sections, as each age group has

lessons for practitioners more commonly exposed to other age groups. The somewhat artificial distinction between child and adult neuropsychology is deliberately blurred in this volume, and hopefully makes for some interesting reading. For example, the child with multiple sclerosis can be compared with the adult with multiple sclerosis, the child with stroke with the adult with stroke, the child with a traumatic brain injury with an adult also injured…there may be valuable lessons about lifespan neuropsychology to be learned from a thorough reading, and that includes perusal of those chapters one might skip if corralled to age-defined sections. We encourage readers to make these leaps into case discussions that do not often come their way.

We wish to thank all of the contributors to this volume for their willingness to join us in this endeavor and for their successful efforts to express their clinical acumen through the case presentation format. We especially want to acknowledge the support and encouragement of Joan Bossert, vice president and editorial director of the Medical Division, Mark O'Malley, production editor, and Aaron van Dorn, editorial assistant, at Oxford University Press. Their support for this project has been fundamental in allowing this volume to be published.

Joel E. Morgan, New Jersey
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Casebook of Clinical Neuropsychology

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Part I

Genetic/Developmental Disorders

Genetic and developmental disorders are many and varied. These disorders are common reasons for referral in neuropsychological practice. They may first present in infancy or by early childhood as the neurocognitive and emotional sequelae of abnormal central nervous system development takes its toll on educational performance. The 13 chapters in this section include both common and rare disorders. These cases are presented to inform both the pediatric and adult neuropsychologist alike, and they illustrate that lifelong challenges are often faced by individuals with these disorders.

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I

Fetal Alcohol Spectrum Disorders

Kimberly Kerns and Heather Carmichael Olson

Fetal alcohol spectrum disorders (FASD) is an umbrella term describing a range of outcomes seen among individuals who are born following prenatal exposure to alcohol. Alcohol is a significant neurobehavioral teratogen, which can cause central nervous system (CNS) damage varying from microcellular and neurochemical aberrations to gross structural anomalies. At the functional level, prenatal alcohol exposure can lead to neurodevelopmental disabilities that range from mild developmental delays or learning disabilities to global cognitive deficits. Fetal alcohol spectrum disorders occur in males and females, among all ethnicities, and across all socioeconomic levels. Reported rates of conditions along the fetal alcohol spectrum vary, depending on the population studied and surveillance methods used, with some calculating the rates of the full range of FASD as high as 9 or 10 per 1000 live births (May & Gossage, 2001; Sampson et al., 1997). Current estimates translate to about 40,000 alcohol-affected births in the United States each year (Lupton, Burd, & Hardwood, 2004).

It is important to accurately identify and understand the full range of neurodevelopmental disabilities arising from the effects of prenatal alcohol exposure, so that appropriate services can be provided to affected individuals. Accurate identification and treatment are needed because many individuals with FASD show significant learning problems and/or maladaptive behavior that prevents them from leading productive, independent lives, and this results in significant societal costs (Burd, Cotsonas-Hassler, Martsolfa, & Kerbeshianb, 2003; Lupton et al., 2004; Stade, Ungar, Stevens, Beyene, & Koren, 2006; Streissguth et al., 2004). Alcohol use during pregnancy, and the issues of offspring born with FASD, are a global public health concern (see http://www.gfmer.ch/Guidelines/Pregnancy_newborn/Fetal_alcohol_syndrome_alcohol_in_pregnancy.htm [accessed 1/10/2009]).

Definitions

The most obvious manifestation of the developmental effects of alcohol is the full fetal alcohol syndrome (FAS). Fetal alcohol syndrome is a permanent birth defect syndrome known to be caused by maternal alcohol consumption during pregnancy. Fetal alcohol syndrome is a medical diagnosis defined by a unique cluster of minor facial anomalies, including short palpebral fissure length, philtrum smoothness, and a thin upper vermillion border (upper lip) (Astley & Clarren, 2001), pre- or postnatal growth deficiency, and CNS dysfunction and/or structural brain abnormalities (IOM, Stratton, Howe, & Battagliam, 1996). The specificity of the FAS facial phenotype to prenatal alcohol exposure supports a clinical judgment that the cognitive and behavioral dysfunction observed among individuals with FAS is due, at least in part, to brain damage caused by a teratogen (Astley, 2004). The U.S. Centers for Disease Control and Prevention (CDC) studies show FAS rates ranging from 0.2 to 1.5 cases per 1000 live births, comparable to other common developmental

disabilities such as Down syndrome or spina bifida (Bertrand, Floyd & Weber, 2005; Mirkes, 2003).

Prenatal alcohol exposure, however, is also known to cause a wider spectrum of adverse functional outcomes, whether or not the characteristic facial features occur. Over the years, clinicians and researchers have given a variety of labels to those who lack some or all of the physical features of FAS, but still have neurobehavioral deficits presumed to be related to prenatal alcohol exposure. Labels include descriptive terms used in research such as "prenatal exposure to alcohol" (PEA) (Mattson & Riley, 1998; Sowell et al., 2008) or "prenatally alcohol-exposed" (PAE) (Rasmussen, Talwar, Loomes & Andrew, 2008), and the outdated term "fetal alcohol effects" (FAE), which should no longer be used in clinical or research settings. To label conditions across the spectrum, the Institute of Medicine uses other terms for diagnostic purposes, which are described later in this chapter (IOM et al., 1996).

Indeed, research advances, including neuroimaging research (MRI, MRS, fMRI) and neuropsychological testing, have clarified that not all individuals affected by prenatal exposure to alcohol display the physical features of FAS (e.g., Astley et al., 2009a-d; Mattson, Riley, Gramling, Delis & Jones, 1998; Riley & McGee, 2005). Research has also suggested that the degree and types of neurobehavioral impairments among individuals with heavy prenatal alcohol exposure do not differ between those with and without physical features of FAS (e.g., Fryer, McGee, Matt, Riley & Mattson, 2007; Mattson et al., 1998). While understanding the relationship between the physical and cognitive characteristics is complex and not fully understood (Astley et al., 2009b), what is clear is that no matter what their physical features, those clinically identified with FASD show neurobehavioral impairments.

Diagnosis

Diagnostic systems for clinical and epidemiologic settings are under intensive development. In 1996, the Institute of Medicine (IOM) defined five conditions along the spectrum with categories of: *(1)* FAS with confirmed prenatal alcohol exposure; *(2)* FAS without confirmed prenatal alcohol exposure; *(3)* partial FAS (pFAS); *(4)* alcohol-related neurodevelopmental disorder (ARND); and *(5)* alcohol-related birth defects (ARBD). At that time, the IOM made recommendations that research data be gathered to allow refinement and validation of the diagnostic system(s). Since then, national guidelines for diagnosis have been and are now being developed around the world. For example, guidelines for diagnosing FAS (only) were developed in the United States (Bertrand et al., 2004). Enhanced gestalt and checklist methods (e.g., Burd, Cotsonas-Hasslera, Martsolfa, & Kerbeshian, 2003; Kable, Coles, & Taddeo, 2007; McGee, Schonfeld, Roebuck-Spencer, Riley, & Mattson, 2008) (some defining FAS/non-FAS only), case-defined diagnostic systems diagnosing across the fetal alcohol spectrum (e.g., Astley, 2004), and systems designed specifically to operationalize the original IOM diagnostic criteria across the spectrum (e.g., Hoyme et al., 2005) have been developed. These are being used in clinical and research settings in the United States and in collaborative international research. In Canada, national guidelines for diagnosing conditions across the fetal alcohol spectrum "have adapted the method of the 4-Digit Diagnostic Code… to identify(ing) domains and severity of impairment or certainty of brain damage," and thus are meant to operationalize the IOM guidelines (Chudley, Conry, Cook, Loock, Rosales, & LeBlanc, 2005, p. 172).

Currently the criteria for a diagnosis of full FAS (only) are comparable across most systems. First, the individual must display facial dysmorphology in three areas: *(1)* short palpebral fissures (eye slits); *(2)* smooth philtrum (the ridges between the nose and lips); and *(3)* thin upper lip. Second, there must be growth deficiency, typically defined as height, weight, or height-weight ratio less than or equal to the 10th percentile. Third, there must be evidence of CNS involvement, such as be a known structural abnormality or CNS dysfunction in three or more domains. Finally, full FAS is typically diagnosed in the context of a confirmed history of prenatal alcohol exposure, but it may be diagnosed when exposure is unknown and the previous criteria are met.

Streissguth and O'Malley (2000), however, argued that diagnosing conditions along the full fetal alcohol spectrum based on facial features is

Rank	Growth deficiency	FAS facial phenotype	CNS damage or dysfunction	Gestational exposure to alcohol
4	***Significant*** Height and weight below 3rd percentile	***Severe*** All 3 features: PFL 2 or more SDs below mean Thin lip: rank 4 or 5 Smooth philtrum: rank 4 or 5	***Definite*** Structural or neurologic evidence	***High risk*** Confirmed exposure to high levels
3	***Moderate*** Height and weight below 10th percentile	***Moderate*** Generally 2 of the 3 features	***Probable*** Significant dysfunction across 3 or more domains	***Some risk*** Confirmed exposure. Level of exposure unknown or less than rank 4
2	***Mild*** Height or weight below 10th percentile	***Mild*** Generally 1 of the 3 features	***Possible*** Evidence of dysfunction, but less than 3 domains	***Unknown*** Exposure not confirmed present or absent
1	***None*** Height and weight at or above 10th percentile	***Absent*** None of the 3 features	***Unlikely*** No structural, neurologic or functional evidence of impairment	***No risk*** Confirmed absence of exposure from conception to birth

Note: PFL = palpebral fissure length; SD = standard deviation. Thin Lip and Philtrum assessed with Philtrum Guide.

Figure 1-1. 4-Digit Diagnostic Code criteria for fetal alcohol spectrum disorders. CNS, central nervous system; FAS, fetal alcohol syndrome. (Figure used by permission of Susan Astley, PhD.)

problematic, especially because the FAS face arises from prenatal exposure occurring during only a very short period of vulnerability in embryonic development, and so is quite tied to the timing of prenatal alcohol exposure (Sulik, Johnston, & Webb, 1981; Sulik, 2005). Chudley and his colleagues (2005) stated that "in the wide array of FASDs, facial dysmorphology is often absent and, in the final analysis, has little importance compared with the impact of prenatal alcohol exposure on brain function" (p. 56). Given this debate, there has been increasing recent diagnostic emphasis on the neurobehavioral deficits presumed to be related to prenatal alcohol exposure, as these are of greater functional significance than the physical features.

It is certainly of clinical, epidemiological, and research interest to generate accurate diagnoses of individuals, and to reliably differentiate between meaningful subgroups on the fetal alcohol spectrum. Consensus has not yet been reached on a single diagnostic system for FASD, and while there are many areas of agreement between the systems in common use, they may sometimes yield different diagnostic classifications when applied to the same alcohol-exposed individual. As data accumulate, diagnostic accuracy will improve.

Diagnostic System Used in Case Studies

The University of Washington Fetal Alcohol Syndrome Diagnostic and Prevention Network (FAS DPN) 4-Digit Diagnostic Code (Astley, 2004; Astley & Clarren, 1997) was used to diagnose the two children in case studies presented in this chapter and so is discussed here in more detail. The 4-Digit Diagnostic Code, now in its third edition, comprises a case-defined set of FASD diagnostic guidelines used by many interdisciplinary teams in the United States and other countries that can be used to define clinical subgroups on the fetal alcohol spectrum. This widely used system aims to reduce classification error. Using this diagnostic system, team members

evaluate evidence for the following: *(1)* confirmed prenatal alcohol exposure; *(2)* level of pre- or postnatal growth deficiency; *(3)* specific facial anomalies characteristic of the FAS facial phenotype; and *(4)* presence of neurostructural anomalies or other "hard" evidence of neurological impairment (e.g., seizures, sensorineural hearing impairment, small head circumference, positive findings on a clinical MRI), *and* the presence, type, magnitude and breadth of neuropsychological deficits across multiple developmental domains. Each diagnostic criterion is evaluated on a four-point Likert scale, assessing the evidence confirming presence of and/or similarity to the presentation seen in the full fetal alcohol syndrome (FAS) and assessing severity. This coding scheme provides a simple yet structured way to capture the complex, variable way dysfunction related to prenatal alcohol exposure is expressed. Using this system, interdisciplinary teams can render an accurate and comprehensive diagnosis of a condition on the fetal alcohol spectrum and provide referrals and treatment recommendations. Teams usually include a physician, psychologist, social worker or public health nurse, clinic coordinator, and some combination of additional members (speech-language pathologist, occupational therapist, family advocate, and other discipline(s) as appropriate). In using the 4-Digit Diagnostic Code process, evidence from neuropsychological testing can (and often does) play a pivotal role in diagnosis and in intervention planning.

Areas of Functional Compromise

Research studies of FASD reveal a variety of primary neuropsychological deficits, quite consistent with the diffuse teratogenic effects expected from prenatal alcohol exposure. Group studies of samples with FASD, compared to control samples, yield testing evidence of lowered IQ, deficits in attention, difficulties in working memory, slowed processing speed, problems with cognitive flexibility, memory deficits, impairment in visual spatial abilities, difficulties in language (especially higher-order integrative language abilities), impairment in motor and sensory skills, and deficits in executive functions (Carmichael Olson, Feldman, Streissguth, Sampson, & Bookstein, 1998; Church & Kaltenbach, 1997; Coggins, Olswang, Olson, & Timler, 2003 Hamilton, Kodituwakku, Sutherland, & Savage, 2003; Jirikowic, Olson, & Kartin, 2008; Lee, Mattson, & Riley, 2004; Kodituwakku, 2007; Mattson, Goodman, Caine, Delis, & Riley, 1999; McGee et al., 2008; Rasmussen, 2005; Thorne, Coggins, Olson, & Astley, 2007).

Beyond a list of functional domains that may be affected by prenatal alcohol exposure, more general research-based statements can be made. Evidence so far suggests that, regardless of overall intellectual level, those with FASD show cognitive deficits at a greater rate than anticipated given their IQ (Kerns, Don, Mateer, & Streissguth, 1997; Schonfeld, Mattson, Lang, Delis, & Riley, 2001). Also, there appears to be considerable *individual* variability within the neuropsychological profiles among those with FASD when wide-ranging test batteries are used (e.g., Astley et al., 2009b; Carmichael Olson et al., 1998). While a growing number of studies suggest there are likely some commonalities in functional compromise, to date an accepted "behavioral phenotype" has not emerged.

Reviewing the body of neuropsychological evidence so far, Kodituwakku (2007) makes a compelling argument that individuals with FASD often have intact performance on simple tasks (in all cognitive domains) but have a "generalized deficit in processing complex information" (p. 199). He argues that reduced intellectual skills and slow information processing are consistent with this generalized deficit. Further, he makes the point that when tasks require integration of multiple brain regions, individuals with FASD are not able to integrate the information needed to meet task demands.

Of further importance among individuals with FASD are significant deficits seen in social and adaptive behavior. These have consistently been noted in clinical literature and systematic research, especially in the areas of communication and social skills (e.g., Jirikowic, Kartin, & Olson, 2008; Jirikowic, Olson & Kartin, 2008; O'Connor, & Paley, 2009; Streissguth et al., 2004; Thomas, Kelly, Mattson, & Riley, 1998; Whaley, O'Connor, & Gunderson, 2001); adaptive behavior as reported by parents is often even lower than what might be anticipated based on overall intellectual ability, at least in the area of social skills (Astley et al., 2009b; Thomas et al., 1998).

Research reporting on data from other informants such as teachers, comparison studies with other disability groups, and information on specific deficits in social skills and social communication need further investigation. As a general statement, it could be said that as situations demand more complex adaptive behavior and social interactions—and so require increased integration of information or place higher demands on executive functioning—alcohol-affected individuals show more difficulty (Coggins, Olswang, Olson, & Timler, 2003; Kodituwakku, 2007; Schonefeld, Paley, Frankel, & O'Connor, 2006; Siklos, 2008).

Clinical studies and systematic research also reveal a wide variety of "secondary disabilities" in lifestyle and daily function among those with FASD, such as disrupted school experiences, trouble with the law, inappropriate sexual behaviors, and more. Most frequent among these secondary disabilities are mental health problems. Research data document a high prevalence of psychiatric conditions and elevated behavior problems among children, adolescents, and adults with FASD (e.g., Mattson & Riley, 1999; O'Connor & Paley 2009; Roebuck, Mattson & Riley, 1999; Schonfeld, Mattson, & Riley, 2005; Spohr, Willms & Steinhausen, 2007; Steinhausen & Spohr, 1998; Streissguth et al., 2004; Streissguth, Barr, Kogan & Bookstein, 1997). However, causal interpretation of deficits within social/emotional and psychiatric domains is usually complicated because this disability group also shows a high prevalence of environmental risks leading to life stress such as early neglect, multiple placements (impacting attachment), abuse history, lack of parental supervision, and parental psychopathology. There are also often genetic/family history factors associated with parent(s) who have possible substance abuse, attention-deficit/hyperactivity disorder (ADHD), learning disorders, or other issues (e.g., Lynch, Coles, Corly, & Falek, 2003). Indeed these factors likely play to some extent into all areas of functional compromise seen in children with FASD (cognitive, adaptive, social, and mental health) and warrant further investigation. Few clinical studies so far have had sufficient statistical power or adequate comparison samples to fully address all these confounding factors, though multiple, well-designed longitudinal prospective studies of prenatal alcohol exposure on offspring development have controlled these variables and confirmed the teratogenic effects of alcohol.

Neuroanatomical and Neuroimaging Findings

There is a growing body of research in the field of FASD confirming permanent anatomical differences in those with FASD on a wide variety of brain structures. In general, findings include greater cortical thickness, smaller brain size, and less white matter density in parietal and posterior temporal regions. Abnormalities have also been noted in the cerebellum, corpus callosum, basal ganglia, hippocampus, and amgydala (Astley et al., 2009a; Riikonen, Salone, Partanen, & Verho, 1999; Sowell et al., 2001, 2002, 2008; Swayze et al., 1997). Variations in cognitive processing as assessed by measures of functional neuroimaging have also been noted. In individuals with FAS, Riikonen et al. (1999) found increased blood supply to the right frontal region, characteristic of children with ADHD. Using functional magnetic resonance imaging (fMRI), a number of authors have found differences in frontal lobe activations in individuals with FASD during tasks of working memory and inhibitory control (Astley et al., 2009d; Connor & Mahurin, 2001; Fryer, McGee, et al., 2007; Malisza et al., 2005). While a comprehensive review of the work in this area is beyond the scope of this chapter, the reader is referred to an excellent review by Spadoni, McGee, Fryer, and Riley (2007). Clearly this work will be important in more specifically elucidating the teratogenic effects of alcohol on brain structure and function.

The Importance of Neuropsychological Assessment and Factors to Consider

While "FASD" is an umbrella term, conditions on the fetal alcohol spectrum are considered medical diagnoses. Current guidelines state that diagnosis is best done within the context of a multidisciplinary or interdisciplinary team. Neuropsychologists can play a unique and important role within these teams, given their training in neuroanatomy and neurology, strong psychometric and assessment skills, and familiarity

with measures used to evaluate multiple domains (language, motor, social-emotional functioning, and sensorimotor). With this background, neuropsychologists can bridge disciplines and bring versatile skills to situations where a full diagnostic team is not available (such as in remote locations), providing a multifaceted assessment and diagnostic perspective. Neuropsychologists also play a role in specifying both an individual's deficits and strengths, which can guide rehabilitation, vocational, educational, and social services. They can also shed light on how neurologic and psychiatric factors interact to impact the behavior of an alcohol-affected individual.

Understanding the developmental impact of alcohol as a teratogen is important when conducting neuropsychological assessment. Prenatal alcohol exposure can vary significantly in terms of quantity and pattern (frequency, variability, and timing) of maternal drinking during pregnancy (Aronson, 1997; Maier & West, 2001; Sood et al., 2001; Streissguth, Barr, & Sampson, 1990). There are factors that modify the impact of the alcohol on the fetus, such as the mother's age, nutritional status, use of other substances, and even genetic factors (Delpisheh, Topping, Reyad, Tang, & Brabin, 2008; Gemma, Vichi, & Testai, 2007; Gilliam & Irtenkauf, 1990; Jacobson, Jacobson, Sokol, Chiodo, & Corobana, 2004; McCarver, Thomasson, Martier, Sokol, & Li, 1997; Stoler, Ryan, & Holmes, 2002). Variation in environmental factors during critical developmental periods will also impact the child's outcome. This explains why significant individual variability in level and pattern of CNS dysfunction occurs among alcohol-exposed individuals.

Because marked variability in potential cognitive and behavioral outcomes is to be expected, neuropsychological assessment and standardized testing must encompass a broad range of neurobehavioral capacities. Documenting a profile of deficits and areas of intact abilities is imperative to understanding a child's unique learning (and behavioral) profile. As is standard practice, neuropsychological test results must be taken together with developmental and family history of risks and protective factors, as well as caregiver and teacher reports of functional cognitive, behavioral, social, and academic strengths and weaknesses. Neuropsychological assessment can provide strong evidence to enable a diagnostic assessment of conditions along the fetal alcohol spectrum—and yield a useful description of function in the alcohol-affected individual with implications for treatment and educational programming. The central importance of neuropsychological assessment is clear in the two case studies that follow.

Case Studies

Provided here are two case studies to illustrate the important fact of the remarkably diverse presentations among individuals with FASD. Both case studies show how a supportive family and appropriate services can result in positive outcomes, even in the face of a child's clear learning and behavioral deficits, and even if a child has experienced high-risk circumstances early in life. The first is a case study of a school-aged child with full FAS, and the second is a case study of an adolescent with ARND. These individuals both have conditions diagnosed on the fetal alcohol spectrum, yet they are different from each other in many ways.

Case 1: A Child with Full Fetal Alcohol Syndrome

Assessment results for Case 1, an 8-year-old male of mixed ethnic ancestry, are from testing obtained as part of a research study as his initial diagnostic testing records were not available. Case 1 was originally seen in an FASD diagnostic clinic at age 5 years, secondary to early developmental and behavioral concerns, and a known history of prenatal alcohol exposure. At that time, he was diagnosed with FAS, with a 4-Digit Diagnostic Code of 3444. The initial (growth) digit of "3" in this child's 4-Digit Code indicates that Case 1 showed moderate growth deficiency, either prenatally or postnatally. The second (facial features) digit of "4" in this child's 4-Digit Code indicates a "severe" level of expression of facial features characteristic of the full FAS (compared to age and Caucasian facial norms: small palpebral fissure lengths, thinned upper lip, smooth philtrum). The third (CNS) digit of "4" in this child's 4-Digit Code indicates there was structural evidence suggesting "definite" CNS damage or dysfunction (very small head size).

The final (alcohol exposure) digit of "4" in the 4-Digit Code indicates confirmed exposure to high levels of alcohol (with an exposure pattern consistent with the medical literature placing the fetus at "high risk," generally high peak blood alcohol concentrations delivered at least weekly in early pregnancy).

Because of early neglect, Case 1 was placed with his current caregivers at 18 months of age. Since then, he has lived in a warm family with multiple siblings. During childhood, Case 1 experienced many protective factors at home and in school. His parents have "reframed" their understanding of their son to appropriately understand Case 1's learning and behavior problems in light of his FAS, provided a stable and developmentally stimulating home, and willingly undertaken behavioral consultation intervention specialized for families raising children with FASD.

Observation and Examination Results. At the time of testing, Case 1 was 8 years, 1 month old with diagnoses of both FAS and ADHD. At the time of testing he was taking Ritalin to treat symptoms of ADHD. Case 1 was qualified for school services under the Health Impaired category and receiving supportive school services, including speech-language therapy, tutoring, and a social skills group, and was placed in a regular classroom.

In interview, Case 1's caregivers described their son as an enjoyable, outgoing, happy boy, who was very helpful with chores and willing to accept direction from adults. However, his mother also described difficulties with Case 1's temper, distractibility, and impulsivity. Behavioral concerns also included physical aggression, apparent lying, difficulties maintaining physical boundaries with peers, and trouble at school. Parent report on the Achenbach System of Empirically Based Assessment, Child Behavior Checklist (ASEBA) (Achenbach & Rescorla, 2001), revealed Internalizing and Externalizing Problem scales both in the clinical range, and scores in the clinical range on the DSM-Oriented scales of Affective Problems, Attention-Deficit/Hyperactivity Problems, and Conduct Problems (above the 97th percentile). Case 1's parents had their son involved in age-appropriate activities (scoring at a remarkably high 92nd percentile on the Activities subscale), but he struggled with social skills and school performance. As a result, his Total Competence score was quite low for his age, at the 7th percentile.

The tester found Case 1 easy to relate to, and a child who wanted to please and be looked upon positively, yet he was highly distractible, worked very fast, and was impulsive during testing. He did persist, even when frustrated, but was often disorganized and inefficient in his approach to tasks. Case 1 was very talkative (with poor articulation). His talkativeness was both helpful (he talked out loud to help himself do better) and a problem (at times talking may have interfered with getting activities done). Case 1 also displayed some odd behaviors during testing, such as laughing inappropriately, making noises, and showing intense periods of excitement.

Case 1's intellectual skills were assessed using the Differential Ability Scales—2nd Edition (DAS-II) (Elliot, 2007). Compared to others his age, his problem-solving skills showed significant variability. His verbal reasoning was estimated as markedly below average, at the 1st percentile. His nonverbal reasoning was in the low range, at the 4th percentile. In contrast, Case 1's spatial reasoning was relatively higher, at the 12th percentile. Importantly, his speed of information processing was solidly average, at the 82nd percentile (though perhaps he traded faster speed for lower accuracy). Even though this variability makes an overall score hard to interpret, Case 1's overall General Conceptual Ability Score was 69, at the 2nd percentile, in the very low range.

Similarly, Case 1 showed striking variability on measures of attention, memory, and executive function. On the Test of Everyday Attention in Children (TEA-Ch) (Manly, Robertson, Anderson, & Nimmo-Smith, 1999), for example, Case 1 demonstrated average skill in sustaining attention on a simple auditory task (~80th percentile). On a visual search task, he maintained accuracy (~80th percentile) but to do so was rather slow (~10th percentile). He had significant difficulty on attention tasks that required mental flexibility, assessed impulsivity, or required divided attention for concurrent completion of a visual and auditory task. On these TEA-Ch measures, his scores ranged from the 12th to below the 1st percentiles.

Compared to other children his age, Case 1's overall learning score on subtests from the Children's Memory Scale (CMS) (Cohen, 1997) was low average (21st percentile). But this overall score fails to capture the discrepancy between his relatively poor visual and better verbal learning and recall skills, and his complex performance pattern. In the verbal domain, on the Word Pairs subtest, he showed a learning rate and total performance in the high average range (63rd and 75th percentiles). He was able to recall this verbal information quite well across both short and longer delays (91st and 75th percentiles, respectively). In striking contrast to his solid, age-appropriate performance with verbal information, Case 1's learning in the nonverbal domain, on the Dot Locations subtest, was low. His learning rate was at the 5th percentile with total performance in the borderline range (9th percentile). However, he did retain and recall nonverbal information he had learned at an average level for age after both short and longer delays (37th and 25th percentiles, respectively).

In the area of executive function, Case 1 was given the Behavioral Assessment of the Dysexecutive Syndrome for Children (BADS-C) (Emslie, Wilson, Burden, Nimmo-Smith, & Wilson, 2003). He scored in the impaired range overall and had clear difficulty remaining organized and flexible when solving unique problems that required him to plan and organize. He scored far below average (<0.2nd percentile band), on several parts of the test that required him to attend to details and organize a number of materials. He also struggled with a task in which, under timed conditions, he had to learn a rule and then change his strategy (and stop himself from responding the original way) when the rule was changed. These findings were very consistent with his caregiver's report of executive function as assessed by the Behavior Rating Inventory of Executive Function (BRIEF) (Gioia, Isquith, Guy, & Kenworthy, 2000), on which Case 1 had multiple elevated scores (especially those assessing problems in behavioral regulation). He also had clinically elevated scores on scales assessing deficits in working memory and the ability to organize his approach to problem solving. Case 1's BRIEF profile suggests that he has a tendency to lose emotional control when flexibility is required or when his routines are interrupted.

Case 1 was also administered the Test of Narrative Language (TNL) (Gillam & Pearson, 2004). This test of integrative language abilities requires that a child understand a spoken story, remember the content, and then retell the story. On the TNL, he performed at the 5th percentile overall, which is consistent with his intellectual skills.

Case 1's mother completed an interview with the Vineland Adaptive Behavior Scale-2nd Edition (VABS-2) (Sparrow, Cicchetti, & Balla, 2005) as a measure of day-to-day functioning, including communication, daily living skills, and socialization. Her ratings revealed that, overall, Case 1 had moderately low performance for age, with an Adaptive Behavior Composite Score at the 6th percentile. Communication skills were rated at the 6th percentile, a moderately low score. Socialization skills were ranked at the 5th percentile, also a moderately low score. In these areas, his behavior was similar to a child of approximately 3–4 years of age, an estimate with which his mother concurred as she reported on her son's day-to-day behavior. Daily living skills, however, were rated slightly higher at the 13th percentile, likely because of Case 1's excellent home environment, where his parents have provided him with extra support and consistent, repeated opportunities for learning activities of daily living.

Case 2: A Child with Alcohol-Related Neurodevelopmental Disorder

Case 2 is a 15-year-old female born to a mother with a history of significant alcohol abuse throughout pregnancy (especially binge drinking), as well as cigarette smoking and possibly some illicit drug use. Case 2 was a healthy full-term infant. At birth, healthcare personnel saw no indication of growth deficiency or facial features suggestive of prenatal alcohol exposure. Case 2 was removed from her birth home due to severe physical abuse in infancy, at about age 9 months, and placed with the family who eventually adopted her, providing a stable and supportive placement.

As a toddler, Case 2 was seen for evaluation because of developmental and behavioral problems, including sleep disturbances, significant temper tantrums, limited social interaction and play skills, aggressive outbursts, and flat affect. Assessment at the time revealed language and motor skills in the average range, though Case 2 met criteria for several psychiatric conditions, including oppositional defiant disorder and reactive attachment disorder. Because of this, she was considered "at risk." Case 2 received intensive early intervention, including therapeutic preschool, and her parents received supportive family counseling and training to handle Case 2's behavior. Case 2's adoptive parents were described as exceptionally motivated and involved in the preschool treatment milieu.

At just about age 5, Case 2 was seen for an FASD diagnostic visit following advocacy by her family. At that time, she was diagnosed with a 4-Digit Code of 1223. The initial (growth) digit of "1" indicated no apparent growth deficiency, and the second (facial features) digit of "2" indicated only mild expression of characteristic facial features. The third (CNS) digit of "2" indicated evidence of some mild to moderate CNS delay and/or dysfunction. This was based on testing indicating primarily impaired adaptive functioning, behavior and social problems (impulsivity and difficulties with peers), unusual sensory sensitivities, mood volatility, and some difficulty with "higher order" language skills for her age, in spite of being quite talkative. Case 2's behavior problems and subtle signs of neurologic impairment and difficulties with skills requiring more complex information processing—not cognitive deficits—were the clearest sign of compromise. The final (alcohol exposure) digit of "3" indicated confirmed prenatal exposure, though not at a level consistent with placing the fetus at "high risk." A 4-Digit Code of 1223 falls within the fetal alcohol spectrum using the 4-Digit Code diagnostic system. Providers might consider this a "milder" form of ARND, and Case 2's condition may have appeared milder (in terms of standardized testing evidence) at the age of 5 years. But her condition was *not* mild in terms of behavioral challenges. In addition, as this case study reveals, the term "mild" did not describe the struggles with academic, problem solving, and social issues encountered by this child and family as time went on.

Case 2 entered the school system with behavioral and learning supports in place, qualified as Health Impaired given the FASD diagnosis. However, actually obtaining an appropriate level of services required parent and health-care provider advocacy. Additional testing by a psychologist using a neuropsychological approach was needed to make the case for services. Given Case 2's complex presentation with "layers" of psychiatric diagnoses in addition to FASD, she continued to receive ongoing psychiatric and support services but, over time, Case 2's medical diagnosis on the fetal alcohol spectrum proved essential to an overall understanding of her issues.

At age 10 years, thorough neuropsychological assessment documented overall intellectual abilities within the average range, though with striking variation within subtests (Wechsler Intelligence Scale for Children–Third Edition, Wechsler, 1996) subtest scores ranging from the 1st to 91st percentile). Case 2's lowest scores were on measures of learning, working memory, and recall (Wide Range Assessment of Memory and Learning (Adams & Sheslow, 1990) and NEPSY (Korkman, Kirk, & Kemp, 1997) subtests), with performance ranging from the borderline impaired to the average range. Case 2 also showed difficulties on measures of planning and organization (e.g., Rey-Osterrieth Complex Figure (Waber & Holmes, 1986). She received low average scores on simple measures of visual spatial/motor skills, with increasing difficulty as tasks became more complex or lengthy. At age 10 years, testing evidence revealed overall adequate language skills on tasks such as phonological processing and single-word reading, but she continued to have difficulty when administered tasks assessing integrative or "higher order" language abilities for her age. This important pattern of doing well on more basic tasks, but struggling when presented with increasingly complex information, was reflected in her academic performance. On the Woodcock-Johnson Tests of Achievement–Revised (WJ-R) (Woodcock, McGrew, & Mather, 1989), she scored above average for age in basic reading skills, yet her scores were low average in reading

comprehension. Consistent with literature so far in the field of FASD, Case 2 had significant difficulty on measures of mathematics, scoring in the impaired range and qualifying for a specific mathematics learning disability. Even though Case 2 was engaging, curious, alert, and charismatic, parent report on the ASEBA continued to reveal a high degree of overall problem behavior. Specific problems included impulsivity, inattentiveness, rigidity, and a concrete problem-solving style, problems with organization, and—most challenging for Case 2 herself and for her family—oppositional and aggressive behavior. By late elementary school, Case 2's mental health diagnoses included posttraumatic stress disorder and generalized anxiety disorder. Her ongoing and complex medication regimen (well supervised by a child psychiatrist) included antidepressants and sleep medications, with changeable mood and anxiety as the main target symptoms.

At age 15 years, Case 2 was again seen for follow-up testing, including neuropsychological assessment. She was administered the Wechsler Intelligence Scale for Children–Fourth Edition (Wechsler, 2003) and scored overall at the 4th percentile, lower than might be expected from earlier assessment. In addition, Case 2 still showed striking variation on individual subtests ranging from solid performance on tasks of verbal comprehension and reasoning (Verbal Comprehension Index = 45th percentile), to significant difficulty with tasks of working memory (Working Memory Index = 0.31st percentile). Subtests that showed the greatest change from earlier testing revealed difficulties now clearly emerging in the areas of working memory and slowed processing speed.

Academic testing revealed reading and writing skills in the average range, reflecting the success of Case 2's own learning efforts. Yet she continued to struggle with mathematics, with poor performance on the Woodcock-Johnson III Tests of Achievement (WJ-III) (Woodcock, McGrew, & Mather, 2001) both Calculation Skills (WJ-III <1st percentile) and Math Reasoning (WJ-III = 5th percentile). Language assessment (Clinical Evaluation of Language Fundamentals–Fourth Edition; Semel, Wing, & Secord, 2003) revealed an interesting pattern of difficulty on the Receptive Language Index (3rd percentile), with no difficulty on the Expressive Language Index (91st percentile).

When given behavior problem questionnaires, Case 2's parents continued to report concerns, including clinically significant difficulties on the Behavior Assessment System for Children–Second Edition (BASC-2; Reynolds & Kamphaus, 2005) such as problems with atypicality, hyperactivity, anxiety, depression, withdrawal, and functional communication. Similarly, parent report on the BRIEF revealed deficits in significantly elevated scores on both the Behavioral Regulation and Metacognitive Indices. In the crucial area of adaptive function (Scales of Independent Behavior–Revised; Bruininks, Woodcock, Weatherman, & Hill, 1996), Case 2's overall broad independence measure was at the "limited" level (1.4th percentile), with moderately serious general maladaptive behaviors requiring some ongoing limited support.

Case 2 is certainly a person with many genetic, prenatal, and environmental risk factors, and with "layers" of diagnosable conditions, including FASD and psychiatric concerns. This, however, is balanced by the remarkable protective factors of Case 2's own zest for life, humor and determination, good work ethic, and a supportive and strong family skilled at advocacy. Ongoing assessment and multimodal interventions addressing her evolving picture of target symptoms have given Case 2 and her family useful coping techniques and accommodations. Among others, interventions in the school years and beyond have included individualized educational programming, home schooling efforts that emphasized life skills training and customized tutoring, behavioral consultation provided to her parents, social skills groups, Special Olympics participation, medication, and more. Case 2 has learned to consistently ask for help from supportive adults and to use good coping strategies.

Discussion

As shown by these case studies, children with FASD and their families face many challenges. Not only have these children been affected by a known teratogen, but they typically have complex postnatal environments. Their parents, who

are frequently foster or adoptive parents, or birth parents in recovery, are often under a great deal of stress. For many individuals with FASD, early testing may reveal only mild (or perhaps moderate) difficulties in development or cognition (as seen with Case 2). However, ongoing and increasing problems in language, social skills, and behavior (adaptive and maladaptive), many times in the context of other mental health concerns, are often observed across the developmental course. Over time, individuals with FASD frequently function at below age expectations in cognitive, linguistic, and behavioral domains, even when they do not display the full FAS. Significant variation is seen in test performance across a wide battery of cognitive domains, with poorer performance on tasks that require higher levels of self-organization or complexity and integration of information. Unfortunately, given this pattern, children with FASD may not be identified or qualified for learning-related services early on. As a result, they have often experienced repeated difficulty and failure and have been identified as children with challenging behavior before receiving a diagnosis on the fetal alcohol spectrum.

Research review and case studies presented here reveal that children with FASD typically score more poorly across multiple developmental domains, but there is as yet no known specific "cognitive profile" of strength or weakness that clearly discriminates those with FASD. This complicates the diagnosis for children without physical features. While Kodituwakku's (2007) suggestion of declining performance with increasing complexity of material across domains may hold promise, this suggestion is not yet useful for diagnosis for two reasons. First, while the idea of increasing complexity fits generally with clinical findings, complexity has not yet been defined as a measurable construct within or across domains, though recent studies have begun the effort to do so (e.g., Aragon et al., 2008). Second, it is possible that a pattern of increasing difficulty with complexity may simply reflect generally diffuse cognitive disability—which is seen in other developmental disorders and therefore not specific to FASD. Clearly, further research is needed to examine methods of operationalizing complexity and to assess its impact and specificity on performance among children with FASD.

There is also considerable research support that children with FASD, as a group, show deficits in executive function and working memory using standardized measures. This finding holds true in group studies but, unfortunately, these areas of deficit are not always found at the level of the individual., Indeed, in a carefully diagnosed sample of 20 children with FAS/pFAS who were administered six well-validated tasks of executive function, surprising results showed that on the majority of these measures participants did *not* consistently score in the impaired range (Astley et al. 2009b).

In conclusion, children and adolescents with FASD have cognitive difficulty across a wide number of domains. Those with FAS (as marked by facial features) are more frequently recognized (and perhaps understood as disabled), more easily diagnosed, and thus more likely to be provided with services and educational supports. Research findings show that appropriate services and caregiver understanding function as protective factors—lowering the odds of the secondary disabilities often associated with prenatal alcohol exposure. This could partly explain the surprising research finding that children with FAS actually show fewer problems in lifestyle and daily function than those on the wider spectrum (Streissguth et al., 2004).

It is unfortunate and remarkable that so many alcohol-affected children still go unrecognized and underserved, often even after being seen by mental health or other providers. While early recognition and diagnosis of FASD has been found to be associated with better outcomes over time, diagnosis at any age is key to more appropriate treatment recommendations and to the anticipatory guidance and long-term planning that is essential for a lifelong neurodevelopmental disability such as FASD. Treatment methods appropriate for FASD are beginning to be systematically tested (e.g., Bertrand, J., 2009), which will better guide future intervention recommendations. A thorough neuropsychological assessment, informed by knowledge of FASD and potential treatments, can be crucial in both diagnosis and in defining and obtaining appropriate family, child, and educational services and supports.

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2

Asperger Disorder

Lauren Kenworthy

Autism is a neurogenetic disorder that is behaviorally defined by the presence of a triad of impairments affecting social abilities, communication skills, and flexibility of interests and behaviors. Current theory states that autism occurs along a spectrum of severity, hence the term *autism spectrum disorders* (ASDs). The ASDs include Asperger's Disorder, autism, and pervasive developmental disorder–not otherwise specified (a category for individuals who do not meet full criteria for autism). Recent estimates of the prevalence of autism, strictly defined, are approximately 3–4 out of every 10,000 individuals (Yeargin-Allsopp, Rice, Karapurkar, Doernberg, Boyle, & Murphy, 2003), but this rate rises considerably when including the entire autism spectrum (Chakrabarti & Fombonne, 2001), with a recent estimate from the Centers for Disease Control and Prevention (2007) indicating that 1 in 150 children have an ASD in the United States. The prevalence of ASDs has increased dramatically in the last decade, with the greatest area of increase among high-functioning (borderline or higher intelligence) children. Therefore, it is common for pediatric neuropsychologists to encounter a high-functioning child on the autism spectrum in their clinical practice.

Among high-functioning children with ASD, the *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revision* (2000) distinguishes Asperger's Disorder from high-functioning autism (HFA) based on the presence of intact language milestones and communication skills. Thus, Asperger's Disorder is thought to reflect impairments in only two aspects of the autism triad: social interactions and flexibility of behavior. The distinction between high-functioning autism and Asperger's Disorder is controversial, however, with many researchers and clinicians arguing that the groups are fundamentally the same. Although some have argued that Asperger's Disorder is associated with nonverbal learning disabilities (Klin, Volkmar, Sparrow, Cicchetti, & Rourke, 1995), a review of neuropsychological findings in ASD (Ozonoff & Griffith, 2000) did not find conclusive evidence of differences in the profiles of individuals with HFA and Asperger syndrome. There are some data that indicate that children with Asperger's Disorder may have higher IQ scores than those with HFA, but even here the evidence is inconclusive. This chapter describes the case of boy who is diagnosed with Asperger's Disorder, but it is also relevant to many children with the diagnosis of high-functioning autism.

Medical complications, particularly seizure disorders, are more common in low- than high-functioning individuals with ASD. Psychiatric comorbidities are very common and most prominently include attention-deficit/hyperactivity disorder (ADHD), depression, and anxiety (Leyfer et al., 2006). Learning disabilities are also commonly comorbid, particularly in the area of written language and reading comprehension. Other commonly affected academic abilities include organization, note-taking, and test-taking.

Although there are psychopharmacological interventions to address secondary symptoms in

ASDs, including the use of selective serotonin reuptake inhibitors, central stimulants, and atypical antipsychotic medications, there are no current medical treatments for the core triad of impairments in autism. The primary intervention strategies available to target the social, language, and flexibility impairments in ASDs are linguistic, behavioral, and cognitive. Because children with high-functioning ASD (HF-ASD) are frequently educated in mainstream environments, interventions are best applied in school settings, and the development of an appropriate school program is typically a fundamental step (Klin & Volkmar, 2000). In addition, there is a great deal of variability within the cognitive profiles of children with HF-ASDs. For these two reasons, a neuropsychological evaluation that delineates cognitive strengths and weaknesses and makes specific educational recommendations regarding classroom placement, necessary accommodations, and special education and therapy needs, can serve as the cornerstone of a treatment plan for a school-age child with HF-ASD. The developmental neuropsychological assessment model described by Jane Holmes Bernstein (2000) is ideally suited to the needs of children with HF-ASD because it emphasizes identifying diagnostic behavioral clusters, or domains, which pose specific risks to the developing child in specific contexts (e.g., elementary school). Delineation of those risks then drives practical recommendations for intervention (Baron, 2000). The following case is presented using this assessment model.

Case Presentation: John

Referral/Background

John is an 8-year-old Caucasian boy in the second grade who was referred following difficulties with socialization and schoolwork. He had previously been diagnosed with a sensory integration disorder, motor delay, and anxiety. John is a boy with many strengths, including precocious math and science abilities. His parents also report that he has a good sense of humor, likes to learn, and is devoted to a few important people in his life. Primary concerns are social and academic. John has a best friend who is a member of a family with whom John's whole family socializes, but John generally has difficulty interpreting social information. In school, John rarely initiates social contact and is generally isolated. He is often distracted and silly when required to work cooperatively with other students. In addition, John has difficulty recognizing his teacher's position of authority and sometimes rudely challenges the teacher's requests or responds in a stubborn, inflexible manner. Academically, as a second grader, John generally works above grade level, but he struggles with written language. He has difficulty with both the physical act of writing and with formulating written language. In addition, he has had general problems completing assignments.

John is the product of a full-term pregnancy, complicated by a maternal kidney infection. Fetal distress was noted during delivery, and John received oxygen at birth. There were no further perinatal complications. John's medical history is remarkable for removal of adenoids at age 3 and for allergy problems and ear infections. John has a history of anxiety and sleep difficulty for which he takes Zoloft and Remeron. Developmental history includes age-appropriate attainment of basic fine and gross motor and language milestones. Development of preacademic skills was precocious: he memorized the alphabet at 18 months. Family history is unremarkable for neurological, cognitive, or psychological problems.

John has been evaluated repeatedly in the past. His parents presented a series of reports for review. A physical therapy evaluation just prior to age 4 showed significant delays in fine motor skills, poor postural control, and problems with sensory modulation. Based on these findings, John was given a diagnosis of sensory integration dysfunction. John was given a comprehensive psychological evaluation at age 5 that revealed a large discrepancy between John's average visual and visual-motor skills and his very superior verbal skills on the Wechsler Preschool and Primary Scale of Intelligence–Revised Edition. John demonstrated exceptional verbal reasoning and vocabulary skills, along with well-developed auditory memory skills and math reasoning skills. John was noted to have great difficulty adjusting to changes in routine and to be argumentative and inflexible at times. Speech and language evaluation revealed average to very

high receptive and expressive language skills. Finally, John was given a math evaluation at age 7. He was found to have very advanced abilities in math, performing above the 99th percentile relative to other children in the first grade.

Evaluation Findings

Evaluation findings are presented here in an integrated format, which includes relevant parent and teacher reports, as well as behavioral observations and test performance, all of which are organized by neuropsychological domain.

General Intellectual Functioning was evaluated using the Wechsler Intelligence Scale for Children–Third Edition (WISC-III) (see Table 2.1). Consistent with his previous evaluation, there is an unusual discrepancy between John's very superior Verbal Comprehension Index and his average Processing Speed and Perceptual Organization Indices. Qualitative observation of John's relatively weaker performance on measures of Processing Speed and Perceptual Organization revealed interference of dysexecutive processes, such as impulsivity and disorganization, as opposed to visual processing deficits. In addition, John struggled with the Picture Arrangement subtest of the WISC-III, due to social problem-solving weaknesses that were also evident on the Comprehension subtest.

Executive Control Functions are a primary area of weakness for John, who struggles in particular with inhibition, organization, working memory, and flexibility. Like many children with executive dysfunction, John's *attention* and problem-solving capabilities are improved with structure. John is able to focus well in a highly structured environment, such as the neuropsychological assessment setting, but he has greater difficulty at home, which is inherently less structured. John was verbally and motorically *disinhibited* during this evaluation. He also made a number of impulsive errors on timed tasks. Anxiety appeared to be driving a number of these impulsive errors. He was very sensitive to the timed aspects of the tests and wanted to complete the task correctly. John's *organization* abilities are weak. Despite his excellent vocabulary, John struggled, for example, when asked to access his verbal lexicon efficiently: his performance was in the average range on verbal fluency tasks, which was highly discrepant with his very superior verbal knowledge. He also struggled to organize visual information, as demonstrated by a highly part-oriented approach to the Rey-Osterrieth Complex Figure, which interfered with his ability to copy and remember it accurately (see Figure 2.1; see also the color figure in the color insert section). John's difficulty in being able to see the gestalt or "whole" of things is in direct contrast to his prodigious ability to process discrete units of information, as demonstrated by his precocious mastery of the alphabet. Both organization and *working memory* deficits make John sensitive to load when processing auditory information. He repeated sentences extremely well but had relative difficulty with paragraph-length stories. However, both parent and teacher working memory ratings are within normal limits (see Table 2.2), indicating that John is able to compensate for these relative weaknesses with his superior memory skills. This was directly observed on the Coding subtest of the WISC-III on which he had difficulty keeping track of symbol–number associations until he memorized these associations.

Flexibility is another area of executive weakness for John, who has difficulty shifting from one task or topic to another, making transitions,

Table 2-1. Wechsler Intelligence Scale for Children–Third Edition

Verbal (Scaled Scores)		Performance (Scaled Scores)		Indices (Standard Scores)	
Information	**19**	Picture Completion	**9**	Verbal IQ	**146**
Similarities	**19**	Coding	**10**	Performance IQ	**95**
Arithmetic	**19**	Picture Arrangement	**6**	Full Scale IQ	**124**
Vocabulary	**19**	Block Design	**13**	Verbal Comprehension	**150**
Comprehension	**13**	Object Assembly	**8**	Perceptual Organization	**94**
		Symbol Search	**10**	Processing Speed	**101**

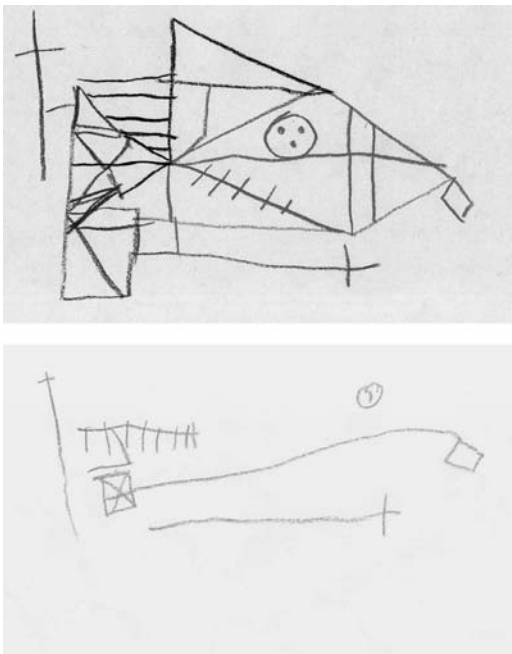

Figure 2-1. (*Top*) Rey-Osterrieth Complex Figure copy condition. (*Bottom*) Rey-Osterrieth Complex Figure delay condition. "See Figure 2.1; see also the color figure in the color insert section."

and who engages in repetitive behaviors, such as pacing obsessively. Thus, although John generally benefits from structure, he has a tendency to become stuck on an idea and is unable to utilize feedback. John demonstrated a number of repetitive routines during testing, including repeatedly using the same phrase. Parent and teacher reports indicate that John can be overly focused and "stubborn" at times, having significant difficulty shifting at school and at home. John was also observed to have difficulty elaborating his answers on portions of testing.

Table 2-2. Behavior Rating Inventory of Executive Functions (T-scores)

Scale/Index	Parent	Teacher
Inhibit	**67**	**56**
Shift	**77**	**79**
Emotional Control	**61**	**71**
Behavior Regulation Index	**70**	**68**
Initiate	**58**	**67**
Working Memory	**50**	**54**
Plan/Organize	**76**	**Missing data**
Organization of Materials	**46**	**64**
Monitor	**79**	**70**
Metacognitive Index	**64**	
Global Executive Composite	**68**	

Language-Related Abilities were striking for the contrast between John's phenomenal command of small units of verbal information, including vocabulary and verbal facts, and his relatively poor ability to hold verbal commands in working memory, organize, or flexibly interpret language. During the evaluation, John was initially relatively quiet, which may have reflected test anxiety. As he warmed up, he became more talkative. Articulation, intonation, volume, rate, vocabulary, syntax, and comprehension were generally within normal limits. Consistent with the previous speech and language evaluation, he scored in the average to superior range on basic measures of language expression (Clinical Evaluation of Language Fundamentals III-Screener standard score = 109) and comprehension (Menyuk Syntactic Comprehension Test standard score = 130). John's verbal knowledge for small chunks of information, such as words and facts, is truly remarkable, as he performed at the ceiling of the Information, Similarities, and Vocabulary subtests of the WISC-III. However, executive dysfunction and weak language pragmatics interfered with his ability to apply his linguistic gifts. A relative weakness was observed when John was asked to use his verbal knowledge to solve problems of daily living on the WISC-III Comprehension subtest. This test required John to process longer and more complex questions than asked on other verbal subtests and to apply his general knowledge to generate common-sense solutions to problems. He also struggled on a timed oral formulation task that required him to produce sentences to describe pictures using two or three specific words or phrases, a laboratory finding that was consistent with his teacher's report that John has difficulty formulating his thoughts on written assignments. John also had difficulty with a semantic flexibility task that required him to generate multiple meanings for words.

Visual-Spatial/Visual-Motor performance on a variety of WISC visual-spatial tasks was age appropriate but represented a relative weakness

compared to John's verbal abilities. His performance was consistently negatively affected by executive dysfunction. Specifically, John made impulsive errors and was penalized for inefficient, disorganized problem solving on timed tests. John's core visual strengths were evident when he was required to work with abstract visual designs on a structured task. It was a pleasure to watch John copy the abstract visual designs presented on the Block Design subtest, for example. Given a model to copy, he worked quickly, observing visual patterns, and replicating them easily. Consistent with his superlative math skills, John enjoyed working with abstract shapes, angles, and lines in space. His high average score underestimates his abilities, as he made multiple impulsive errors on this task. John had greater difficulty on visual problem-solving tasks that required perception of visual gestalts, organization of visual information, or social problem solving. For example, he had great difficulty assembling a puzzle of a human face.

John's visual-motor capacities have improved since his motor evaluation at 4 years of age. He has benefited from occupational therapy and holds his pencil with an appropriate grasp. John is able to provide appropriate fine motor output over short periods of time, achieving, for example, an average score on the WISC-III Coding subtest. However, ongoing evidence of weakness in this area is provided by his poor bimanual control of pencil and paper and poorly controlled drawing (see Figure 2-1) and letter formation in writing samples. Both teacher and parent report ongoing difficulty with sustained writing. Slowed motor response time was evident on the Test of Variables of Attention, a continuous performance test. Thus, motor difficulties interfered with visual-motor coordination, particularly over time. Executive dysfunction, however, was the primary impediment on visual-motor tasks, as evidenced by the almost two standard deviation drop between his high average performance (standard score = 110) on the structured, relatively simple Beery-Buktenica Developmental Test of Visual-Motor Integration and his low average (standard score = 84) score on his copy of the Rey-Osterrieth Complex Figure. As described earlier, John took an extremely part-oriented approach to the Rey and failed to integrate its component parts.

Learning and Memory was excellent for small units of verbal or visual information, but executive dysfunction interfered with John's ability to learn larger chunks of information and retrieve information from memory. He performed above age expectations in his ability to learn small units of visual or verbal information (Wide Range Assessment of Memory and Learning [WRAML] Sentence Memory standard score = 17; Children's Memory Scale [CMS] Dot Locations scaled score = 15; CMS Number Span scaled scores: Forward = 18, Backward = 19). However, his performance deteriorated as the complexity of information increased due to interference from executive dysfunction (WRAML Story Memory scaled score = 12; Rey-Osterrieth Complex Figure Delay Memory Inventory standard score = 74). John is able to maintain small units of numbers, sentences, and visual data by utilizing his remarkable memory span. He also does very well when given the opportunity to memorize larger chunks of data. For example, on a list-learning task, John was initially overloaded by the large amount of data presented to him, as demonstrated by a major primacy effect in his recall of the list. However, with repetition he was able to compensate with strong rote memorization skills and perform at an above-average level (see CVLT scores below).

California Verbal Learning Test-Children's Version (standard scores)

Total Score **117** Trial 1 **108** Trial 5 **123** Learning Slope **123** Recall Consistency **123** Short Delay Recall Free **100** cued **115** Long Delay Recall Free **123** cued **108** Discriminability **123** (Recognition) Semantic Cluster **77** Primacy **115** Recency **77** Perseverations **92** Interference **100**

John demonstrates weaknesses in the area of *social cognition and social skills*. Social cognition is the ability to make sense of socially relevant information and feelings. It includes interpretation of nonverbal cues, understanding of human relationships, and social problem solving. It also addresses expressive abilities to communicate feelings through nonverbal cues, social motivation, and theory of mind. As noted above, John had difficulty on WISC-III subtests requiring social cognition and even commented after the Picture Arrangement subtest, "I hated that."

Related to this, John had a great deal of difficulty with the Roberts Apperception Test, which required him to tell brief stories describing line drawings of people experiencing emotions or interacting socially. Moreover, he was unable to respond to multiple choice questions about what people in the pictures might be thinking or feeling (e.g., Is this person happy, sad, mad, or scared?). John had great difficulty when describing his own feelings, as well as describing important characteristics of people in his family and human relationships. For example, when asked, "Why do people get married?" his response was "so they can reproduce." During open-ended questions, it was clear that although John wants to have friends, he is more focused on, and motivated by, scientific and mathematical concepts than social interactions.

John's social skills reflect his social cognition profile. Like many children on the autism spectrum, John's best relationships are with the most familiar people in his life: his family. He completed the sentence "I love…" with "…my mommy." Associated with this is his early bonding history, including the development of a social smile, stranger anxiety, recognition of his parents after an absence, physical responsiveness, and engagement in basic interaction games (e.g., peek-a-boo), all of which occurred at appropriate ages. At the same time, his misinterpretation of social information, combined with executive dysfunction, creates significant social difficulties. John is reported to be quite withdrawn both at home and at school. In the one-to-one, highly structured testing situation, John presented as an attractive, cooperative, friendly boy. He has a nice smile, was able to make good eye contact with the examiner, and was good at imitating the examiner's intonation. John had difficulty recognizing socially inappropriate behavior, however. He appeared to be unaware of the impact of his actions on others, not recognizing that imitation of the examiner was inappropriate, for example. The prolonged social interaction required by the assessment was stressful for John, and he responded at times with silliness, raising his voice to answer questions, or answering in pig Latin while sitting underneath the table. John also was unable to pick up on conversational cues from the examiner and respond appropriately, a sign of limited social reciprocity.

Emotional Adjustment is generally appropriate, but John's parents describe a history of anxiety and sleep difficulties. John, like many bright children who struggle with executive dysfunction and social learning problems, was somewhat anxious during this evaluation. Of more concern at the present time are both parent and teacher report on the Behavior Assessment System for Children rating scales of increased dysphoria combined with mild anxiety.

Impression

John's neuropsychological profile is characterized by the following: very superior verbal intelligence; executive dysfunction, including weakness in inhibition, organization, working memory, and flexibility; a social learning disorder; and slowed and poorly controlled motor output. This profile is consistent with, although distinct from, the diagnosis of Asperger Disorder. John meets diagnostic criteria for Asperger Disorder by virtue of his impaired social interaction skills, most notably poor social reciprocity and limited peer interactions; subservience to repetitive behaviors/interests, for example, his repetitive pacing, overfocus on math concepts to the detriment of conversations with others, and extreme rigidity regarding routines; intact early language development and current core language abilities; and lack of general cognitive impairment.

John is a child with remarkable *strengths*. He truly enjoys learning. His performances on measures of vocabulary, verbal reasoning, factual knowledge, and mental arithmetic are at the ceiling of our ability to measure them. He has a powerful memory span, which allows him to memorize small units of information with facility. He can excel at learning larger chunks of information as well, as long as it is presented repeatedly. Moreover, John has superior mathematical and scientific abilities and makes excellent use of his knowledge to support high-level strategic thinking. His ability to think strategically is a prognosticator of his future ability to compensate for areas of difficulty. In fact, John already demonstrates that he makes remarkable compensatory efforts. When John is explicitly taught information that he does not intuitively grasp though rote learning using rules, recipes,

and routines, it is predicted that he will make great strides.

John's neuropsychological protocol puts him at *risk* in a variety of situations and with a variety of tasks:

1. John is at risk for being misunderstood and overloaded. His remarkable vocabulary, abstract reasoning, fund of knowledge, and mathematical and scientific abilities are difficult to reconcile with his difficulty completing tasks, learning cooperatively in groups, organizing his thoughts coherently, and flexibility responding to the requests of others. These discrepancies are most readily explained through assumptions of stubborn behavior or inadequate effort on John's part. Moreover, as is the case with many extremely bright children such as John, his compensatory abilities are remarkable and have in many situations masked core deficits. For example, John's remarkable facility at processing words and sentences will lead people to assume that he is a much stronger auditory processor of longer chunks of information than is the case, and to overload him with information. When overwhelmed, the combination of cognitive weakness and anxiety can produce a severely inflexible child.
2. John is also at risk for difficulty with information output. John has difficulty producing work in the classroom because disinhibition, weak working memory, disorganization, and inflexible routines all conspire to limit his ability to work independently. Furthermore, although he has a great store of verbal facts and details, this is not matched by his ability to formulate language and organize ideas. This places John at great risk for frustration over his inability to communicate his ideas. Moreover, while improved, John's long-standing fine motor difficulties continue to hinder his handwriting, which adds a considerable load whenever John is asked to put pencil to paper and thus slows his output.
3. John is susceptible to overfocusing on literal details of instructions and extensive reliance on verbally based, rotely learned rules and approaches to learning. Although this approach may support academic development and is very useful to John in a variety of settings, it may also interfere at times with creative problem solving. More importantly, this style, combined with biologically based inflexibility, may result in apparently rigid behavior when John may simply be having trouble taking in the larger metaphor or meaning of a situation. This thinking style puts John at risk for academic difficulty with expository writing and reading comprehension.
4. John is at high risk for boredom unless he is exposed to a demanding curriculum in math and science.
5. John's difficulty accurately interpreting social information and his constitutional inflexibility places him at risk in his interactions with his teachers and peers. He is likely to miss important information about the expectations of others and get stuck with his own ideas and ways of doing things. Thus, he may not respond appropriately to feedback. He is at particular risk in peer group interactions where he will have the most difficulty controlling the agenda and limiting the amount of information he must process simultaneously. His weakness in organization makes it particularly difficult for him to integrate the information that he receives in such settings. Thus, cooperative learning tasks are very challenging for a child like John.

Recommendations

Recommendations for John use his strengths to combat these risks. They focus on the following: the appropriate school placement for John; the individual therapies and special education supports that he requires; and the accommodations he needs in school. John will require some specialized and individualized services at school; therefore, in a public school setting he must be coded for special education services. The findings of this assessment support coding John as a child with an ASD who is also intellectually gifted. His intellectual abilities make him appropriate for a mainstream classroom if he receives specialized supports.

In order to learn and produce information in cooperative group settings, follow teacher instruction appropriately in class, and maintain appropriate peer relationships, John requires the attention of *a specialized team to support*

social development. If appropriately trained, a school psychologist and speech and language therapist can work together to provide counseling, behavior management, and training in social skills and the pragmatics of communication. John requires a "social coach," an adult who genuinely likes and understands him, who can help him learn to improve his ability to pick up on concrete indicators of the feelings of others (e.g., when somebody stands that way, it often means he is angry); logically deconstruct the meaning of social interactions and learn to take the perspective of others (e.g., how do you think he felt when you told him that his math question was "trivial?"); understand the impact of his own actions on others through explicit, concrete, cause–and-effect explanations; and apply social stories and learn associated social rules. Respect John's learning strengths by teaching him in small increments and providing him with discrete social rules, logical and sequential explanations of social events, and routines that he can memorize. Make social skills training intervention an integral part of all activities implemented consistently across the school setting through the use of a behavioral reinforcement system that rewards prosocial behaviors in all children.

John continues to require *occupational therapy* intervention to assist him in becoming comfortable with a keyboard. In addition, John requires helps with fine motor self-care tasks, such as fastening fasteners.

John requires individualized *special education* to improve his written language expression, including teaching him a specific routine for producing written work that is practiced repeatedly and written down in a checklist (e.g.: 1. Brainstorm ideas; 2. Select appropriate ideas for topic matter and length; 3. Put ideas in order, etc.…). John also requires special education to improve his executive abilities for working independently and completing tasks. In addition to the use of rules, recipes, routines, and checklists, John needs explicit teaching in order to learn skills that other children may learn on their own. Place specific emphasis on approaches for breaking tasks down into small units, and techniques for identifying the main requirement of an assignment. Provide explicit coaching in strategic planning and goal-oriented problem solving, with an emphasis on consistent routines that can be applied to a variety of independent projects. To be effective, coaches teaching new strategies and routines for tackling work, or demands of daily living, must first model the desired routine and then provide extensive practice with a gradual shift from externally cueing John to having him follow internal cues and written rules or routines.

John's academic and social performance will be improved with familiarity and structure, which can be created through the following *classroom-based accommodations*. Provide John with regularly scheduled down time away from the omnipresent social/executive demands of a standard classroom. Give John free access to his social coach throughout the school day if he needs to address a problem. Provide extra structure for any new experiences or learning, including posting a visual schedule; previewing transitions or unexpected events; providing highly structured routines and frequent one-to-one check-ins to structure independent activities; and using outlines, worksheets, checklists, recipes, written routines, and other interventions to teach John in a step-by-step fashion. Keep oral directions brief or accompany them with a visual reminder, such as a checklist. Take every opportunity to write directions down for John to provide him with visual cues regarding steps he needs to take to carry out work independently. Eliminate handwriting requirements through use of a keyboard or voice-activated software. Apply testing accommodations. Support organizational deficits: Review homework assignments with John before he leaves school each day; assist with the maintenance and organization of a notebook; and provide a daily exchange of information between home and school about assignments and goals.

Finally John's family was encouraged to *engage a psychologist outside of school* to monitor and provide cognitive-behavioral treatment for John's anxiety, in addition to developing a positive behavioral management program at home to target increased flexibility and prosocial behaviors.

Epilogue

John was re-evaluated on two subsequent occasions, when he was 11 and 14 years old. At 11, the presenting problems remained social isolation

from peers, inflexibility in conversation and behavior, and poor written expression. His neuropsychological profile was generally consistent with his initial evaluation, although his Verbal Comprehension Index score dropped to 128 on the WISC-IV, while the addition of the Matrix Reasoning subtest to the Performance scale provided him with an opportunity to demonstrate his remarkable visual problem solving in the absence of executive function demands (scaled score = 18). Thus, his overall IQ profile was even, with superior performance in both verbal and nonverbal domains, but there was considerable subtest scatter related to the relative executive demands of the task. John remembered the Block Design subtest from the evaluation conducted almost 4 years previously and requested it when he entered the room for re-evaluation. He continued to be fascinated by and obsessed with mathematical problems. In a further indication of his inflexibility, he appeared for his February evaluation in shorts, because he had been unable to make the transition from fall to winter clothing. John had also developed increased aggressive behavior at home, primarily related to inflexibility.

John's ability to respond to social stimuli had improved, such that he was now able to provide brief stories for the Roberts Apperception cards, but he tended to be very concrete in his responses and often indicated that he did not think the people in the pictures were feeling anything. For example, a picture of a child striding across the floor in anger with a chair over his head was described as "a man asked his son to bring him a chair. His son went to get the chair and carried it back high over his head." When asked explicitly what the person might be thinking or feeling, John said, "not feeling anything substantial."

John's written language remained a major obstacle at school, where he had received handwriting accommodations but no support for the organization of written expression. Conflict between John, his parents, and his teachers had increased, with his teachers insisting that his refusal to write paragraphs was volitional and his parents disagreeing. His performance on a test of written language was discrepant with his Verbal IQ score, and he was given an additional diagnosis of a learning disability affecting written language. Recommendations included the following: a meeting between the evaluator, John's family, and school personnel to problem solve how to teach John to write; behavior management to target aggressive outbursts; and consideration of stimulant medication.

At 14, John's profile was again highly consistent with previous evaluations. He entered the examination room discussing factorials, which he proceeded to introduce into the conversation whenever it was not structured by the examiner. When presented with the Rey-Osterrieth Complex Figure, he immediately remembered it and a feedback session that occurred when he was 11 in which the effective strategy for copying it was discussed. He described the strategy in some detail while he copied the figure in an entirely part-oriented fashion, directly contradicting what he was saying as he worked. He was fascinated by the strategic possibilities of the Tower of London-Dx, but was unable to hold the two rules governing completion of the tower in mind as he worked. His impulsivity continued to hinder his problem solving.

Although John was now taking Strattera in addition to his other medications, and his family was receiving excellent in-home behavioral management support, he continued to have aggressive, impulsive outbursts, particularly in response to flexibility requirements. He had resisted the help of psychotherapists, firing several. Adaptive daily living and socialization abilities are severely impaired by parent report.

John is placed in a high school for gifted and talented children, which readily accommodates his academic needs, and his writing has improved substantially. He is now producing long works, notably a remarkably insightful autobiography, in which he reported difficulty remembering people's names and noted that he was not connecting with other kids in class the way they were connecting with each other. He is valued by his peers for his remarkable math and science abilities, but he is friendless and isolated during unstructured times. A school observation revealed him to spend his lunch period alone in a darkened classroom turning in a revolving chair.

Recommendations centered on identifying specific goals that would enable John to function independently as he approached adulthood: the ability to make a friend, carry out basic daily

living skills, and shape his behavior in response to the requests of others (improve flexibility). Regarding the latter, the examiner recommended explicit cognitive/behavioral training in what flexibility is, what the advantages of being flexible are, and how to be flexible in different situations. A meeting of his treatment team to coordinate these goals and to explicitly pursue John's "buy in" or commitment to pursing these goals was suggested.

Discussion

John is in many ways prototypical of the subset of very high-functioning children with ASD. His profile of poor executive organization and flexibility, social learning impairment, and fine motor weakness, occurring in the context of excellent learning and memory for small chunks of information, is typical of his diagnostic group. Written language learning disabilities, such as those that John has, are also very common. The presence of significant problems with impulsivity is not prototypical, but is common in high-functioning children with ASD. John's difficulty with flexibility is somewhat more severe than is typical, and it may be exacerbated by his remarkable intelligence, which frequently supports his belief that he is smarter than other people. The lack of a family history of an ASD, social learning disorder, or language disorder was unexpected, although subsequent to his first evaluation, John's sister was diagnosed with pervasive developmental disorder–not otherwise specified.

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3

The Identification of Autism Spectrum Disorders in Early Childhood: A Case Report

Marianne Barton, Katelin Carr, Lauren Herlihy, Kelley Knoch, and Deborah Fein

The well-documented benefits of early identification of autism spectrum disorders (ASDs) (for review, see Dumont-Mathieu & Fein, 2005) have led to increased focus on the *earliest manifestations of the disorder*, *the stability/validity of early diagnosis*, and the *identification of assessment tools suitable for use with very young children*. Equally important, recent recommendations for widespread screening of young children for ASDs (AAP, 2006, 2008) have prompted increased interest in *population-based screening measures* and increased pressure to identify children at risk for the disorder as early as late infancy. All of these efforts have been helpful in permitting the ever-earlier identification and treatment of children with an ASD. At the same time, they highlight the complexity and heterogeneity of the disorder, its interaction with normative variation in developmental trajectories, and our limited understanding of early social and communicative development and its aberrations. We will first discuss some of the literature on these issues and then present a case that illustrates many of them.

A variety of *screening measures* are currently in use or development, including the Checklist for Autism in Toddlers (CHAT; Baron-Cohen, Allen, & Gillberg, 1992), the Modified Checklist for Autism in Toddlers (M-CHAT; Robins, Fein, Barton, & Green, 2001), the Pervasive Developmental Disorders Screening Test-II (PDDST-II; Siegel, 2004), Screening Tool for Autism in Two-Year-Olds (STAT; Stone, Coonrod, & Ousley, 2000), the Checklist for Autism in Toddlers-23 (CHAT-23; Wong et al., 2004)), the Early Screening for Autistic Traits (ESAT; Swinkels et al., 2006, and, most recently, the Infant-Toddler Checklist (ITC; Wetherby & Prizant, 2002). The PDDST-II and the STAT have shown good sensitivity and specificity as stage 2 screeners for use with children in developmental clinics; the authors of the PDDST II have reported good specificity and sensitivity as a stage 1 screener as well (Siegel, 2004), but these data have not yet been replicated in a large community sample (Dumont-Mathieu & Fein, 2005). The STAT is a level-two screen designed to differentiate toddlers with autism from those with other developmental disabilities (Stone et al., 2000; Stone, Coonrod, Turner, & Pozdol, 2004). The validation sample for this measure reported a sensitivity of .83 and a specificity of .86 (Stone et al., 2004).

The CHAT was the first level-one, autism-specific screener and consists of five observation items and nine parent-report items (Baron-Cohen et al., 1992; Baron-Cohen et al., 1996). This measure is currently under revision by the authors due to limited sensitivity (20%–38%) found on follow-up (Baird et al., 2000). The ESAT is a screener for autism in children 14–15 months developed in the Netherlands. The 14-item parent-report questionnaire was reported by the authors to have greater than 90% sensitivity for an ASD but poor specificity for differentiating ASDs from other developmental disorders, a finding that may be related to the fact that the ESAT was tested with especially young children (Swinkels et al., 2006).

Among level-one screeners, the M-CHAT and the ITC currently appear to be most promising. The M-CHAT is a 23-item checklist that asks a parent to provide yes/no responses to questions about his/her toddler's development (Robins et al., 2001). It was adapted from the CHAT by removing the pediatrician observation section and adding parent-report items. The authors report good estimates of specificity and sensitivity, although a follow-up interview was added for children who failed the initial screen to reduce the false-positive rate. Positive predictive value for the M-CHAT has been reported as .36 before and .68–.74 after the follow-up interview (Robins et al., 2001; Kleinman, Robins, et al., 2008). The ITC, a section of the Communication and Symbolic Behavior Scales Developmental Profile (CSBS) has recently been examined as a broad-band screener for ASDs (Wetherby et al., 2008). The ITC is composed of 24 items with three to five response choices for questions about social communication milestones, and an additional open-ended question about the parent's developmental concerns (Wetherby et al., 2008). Unique among the screeners reviewed here, the ITC includes standard scores for each month from 6 to 24 based on a large normative sample in addition to a screening cut-off score. The authors report positive predictive values above 70% for children age 9–24 months for communication delays, and 93.3% sensitivity for ASDs in particular, although they caution that a positive screen on the ITC does not distinguish these two groups (Wetherby et al., 2008).

Once children are identified as being at risk for an ASD, they are referred for a more detailed diagnostic evaluation.

In contrast to earlier assertions, it is now clear that *valid and stable diagnoses* of an ASD can be accurately made in children under the age of 3 (Charman et al., 2005, Cox et al., 1999, Eaves & Ho, 2004; Lord, 1995; Moore & Goodson, 2003; Stone et al., 1999). The accuracy of early diagnosis has been further supported by studies investigating the stability of diagnoses given between the ages of 2–3 years with confirmatory diagnoses given 1, 2, and 3 years later (Chawarska, Klin, Paul, & Volkmar, 2007; Cox et al., 1999; Eaves & Ho, 2004; Kleinman et al., 2008; Lord, Storoschuk, Rutter, & Pickles, 1993; Moore & Goodson, 2004; Turner & Stone, 2007). While overall, ASD diagnoses are considered relatively stable, diagnoses of autism (72%) are more stable than diagnoses of PDD-NOS (42%) (Stone et al., 1999). It is unclear whether the limited diagnostic stability in individuals diagnosed with PDD-NOS at age 2 years is due to higher responsiveness to treatment, the ambiguity in the diagnostic category, or a tendency for the milder symptoms to be transient (Cox et al., 1999; Eaves & Ho, 2004; Stone et al., 1999; Walker et al., 2004).

Some of the earliest manifestations of ASDs have been fruitfully studied with early home movies of diagnosed children, as well as prospective studies of high-risk children (i.e., younger siblings of affected children). These studies have revealed that early signs of autism vary appreciably with age during the period from infancy through early childhood. During the first year of life, children with an ASD are less likely than their typically developing counterparts to orient when their name is called (Osterling & Dawson, 1994; Osterling, Dawson, & Munson, 2002; Werner, Dawson, Osterling, & Dinno, 2000). An absence of social smiling and lack of facial expression have also been identified as early markers of an ASD (Adrien et al., 1992; Zwaigenbaum et al., 2005). Infants with an ASD look at others less frequently than typically developing infants (Osterling & Dawson, 1994; Osterling et al., 2002) show deficits in visual tracking and imitation (Osterling et al., 2002; Zwaigenbaum et al., 2005), and they show fewer social and joint attention behaviors (Osterling & Dawson, 1994; Zwaigenbaum et al., 2005). In the second year of life, children with an ASD show an increased lack of response to name and poor eye contact (Chawarska et al., 2007; Wetherby et al., 2004), as well as failure to point (Chawarska et al., 2007; Cox et al., 1999), delays in functional and symbolic play, and limited response to joint attention prompts (Chawarska et al., 2007). Deficits in sharing enjoyment and interest, lack of facial expression, and lack of showing also characterize this population (Wetherby et al., 2004). Some researchers have found increased frequency of repetitive behaviors in the second year (Wetherby et al., 2004), although others have not (Cox et al., 1999).

Changes in symptom presentation in children with an ASD between the second and third years of life include the emergence of speech

(Charman et al., 2005; Chawarska et al., 2007), in addition to the acquisition of atypical language features, such as echolalia and unusual intonation (Chawarska et al., 2007). Increased bids for joint attention, a lack of pointing, and a marginal increase in communicative gesture use have also been found in children with an ASD during this period, as well as a lack of improvement in coordination of social-communicative behaviors, eye contact, and direction of facial expression toward others (Chawarska et al., 2007).

In the fourth year of life, Cox et al. (1999) found that children with autism continue to exhibit reduced affect sharing and imaginative play relative to children with language disorder. In Charman et al.'s (2005) longitudinal study of children with an ASD, children gained more reciprocal social interaction skills between the ages of 4–5 and 7 years, whereas gains between 3 and 4–5 years were not significant. However, significant gains in communication skills were made in Charman et al.'s (2005) sample from ages 3 through 7.

Communication and social interaction deficits appear to be the most salient characteristics of young children with an ASD, whereas repetitive and stereotyped behaviors in young children have been less fully understood. Repetitive behaviors may be less frequent or less noticeable at younger ages and increase by 42 months (Cox et al., 1999), but other researchers report no changes in the level of repetitive behaviors between the second and third year of life (Charman et al., 2005; Chawarska et al., 2007). Children with an ASD younger than 36 months have been found to exhibit more simplistic repetitive behaviors, such as hand and finger mannerisms and repetitive use of objects, whereas children older than 36 months appear to demonstrate more higher level behaviors, such as resistance to change and circumscribed interests (Mooney, Gray, & Tonge, 2006). Charman et al. (2005) found that repetitive behaviors increased most between the ages of 3 and 4–5 years and then decreased at 7 years. However, Charman et al. (2005) also found significant individual variation in the frequency of repetitive behaviors and other autism symptoms over time.

Variation in the presentation of symptoms of ASDs during early childhood has important implications for the selection of *assessment tools*. Autism-specific assessment tools currently available to clinicians include measures based on caregiver report, such as the Autism Diagnostic Interview-Revised (ADI-R; Rutter, LeCouteur, & Lord, 1995), and those based on the clinician's behavioral observations of the child, including the Autism Diagnostic Observation Schedule (ADOS; Lord, Rutter, DiLavore, & Risi, 2000) and the Childhood Autism Rating Scale (CARS; Schopler, Reichler, & Renner, 1980). These measures must be supplemented by assessment of developmental level, adaptive skills, and communication skills in order to provide a context in which to evaluate the meaning of atypical social behaviors.

Clinical judgment is considered the "gold standard" for diagnosing ASDs in children less than 5 years old (Volkmar et al, 2005). Both clinicians and the diagnostic measures available rely on criteria from the *Diagnostic and Statistical Manual for Mental Disorders-IV* (*DSM-IV*). Stone and colleagues (1999) have studied the applicability of *DSM-IV* criteria to young children with autism. Their study found the criteria related to social deficits and delayed language to be most prominent in children under 3 years of age. However, other items, such as failure to develop peer relationships, impaired conversational ability, stereotyped and repetitive use of language, and inflexible adherence to routines and rituals, were not as reliably observed in young children with autism.

The stability of diagnoses has been explored based on clinical judgment and commonly used diagnostic instruments. In a study by Kleinman, Ventola, et al. (2008), the initial diagnosis of 77 children (mean age of 2.25 years) was compared with the confirmatory diagnosis made 2 years later (mean age 4.4 years). The authors report high stability for clinical judgment based on the *DSM-IV* (80%), with higher stability for diagnoses of autistic disorder (70%) compared to PDD-NOS (33%). The stability rate for diagnosis was at acceptable levels for diagnosing ASDs in toddlers, with only 15 children moving off the autism spectrum at follow-up and none moving onto the spectrum. Stability was high for diagnosis based on the ADOS (83%) and the CARS (76%), but lower for the ADI-R (67%). Unlike the ADOS and the CARS, the ADI requires the presence of repetitive behaviors or restricted

interests to make the diagnosis of autistic disorder. At follow-up, several children who did not originally meet the criteria of repetitive behaviors required for a diagnosis of autistic disorder on the ADI subsequently met criteria after developing restricted interests or repetitive behaviors at age 4. Other studies have also found decreased stability in the ADI-R in 2-year-olds, in part because some children did not develop restricted interests or repetitive behaviors until later in development (Charman et al., 2005; Cox et al., 1999; Turner et al, 2006; Turner & Stone, 2007).

The interrater reliability of diagnostic instruments has also been examined in the early diagnosis of ASDs. When classifying toddlers as spectrum versus nonspectrum, interrater reliability for diagnosis based on clinical judgment, CARS, and *DSM-IV* was considered good, with clinicians agreeing on the diagnosis of 57 out of 65 children (88%). This level of agreement dropped to 64% when distinguishing between autism and PDD-NOS (Stone et al., 1999). High agreement has also been found between clinical judgment based on *DSM-IV* criteria, CARS, and the ADOS-G in a sample of children aged 16–31 months (Ventola et al., 2006). Upon examination of the ADI-R, researchers have found low levels of agreement between the repetitive behaviors domain and other measures (Cox et al., 1999; Saemundsen, Magnusson, Smari, & Sigurdardottir, 2003; Ventola et al., 2006). These studies suggest that a change in the criteria of the ADI-R, namely a decreased cut-off score for the repetitive behaviors and restricted interests domain, may be necessary for the ADI-R to be used reliably in the early diagnosis of ASDs.

The sensitivity and specificity of the diagnostic instruments have been compared to the "gold standard" of clinical judgment. The ADOS-G and CARS have been found to have good sensitivity rates and adequate specificity rates for diagnoses of autism and PDD-NOS (Cox et al, 1999; Ventola et al., 2006). The ADI-R also had adequate specificity rates but relatively poor sensitivity. In one study the ADI-R failed to identify almost half of the children (17 of 36) diagnosed as having autism or PDD-NOS, primarily because these children did not exhibit early repetitive behaviors (Chawarska et al., 2007; Cox et al., 1999; Ventola et al., 2006). Wiggins and Robins (2008) were able to increase the sensitivity rate of the ADI-R, from .33 to .79, with minimal compromise in specificity (.94 to .78) when they excluded the behavioral domain from their analyses.

In addition to consideration of limitations of existing measures for ASD diagnosis, it is important to identify more general developmental concerns related to the assessment of ASDs in toddlers. As noted earlier, there is divergence of autism symptoms from typical behavior changes over the course of early development (Vig & Jedrysek, 1999). Therefore, developmental level or mental age must be considered when interpreting potential autism-related behaviors (Vig & Jedrysek, 1999). Several authors have noted that it is difficult to differentiate global developmental delay (GDD/mental retardation) from ASDs in children with mental ages below 18–24 months (Rutter & Schopler, 1987; Vig & Jedrysek, 1999). The Childhood Autism Rating Scale (CARS) and the Autism Diagnostic Interview-Revised (ADI-R) have been shown to overidentify 2-year-olds and older nonverbal children with mental ages below 18 months (DiLavore, Lord, & Rutter, 1995; Lord et al., 1993; Saemundsen et al., 2003; Vig & Jedrysek, 1999). Specific criteria for ASDs that reference skills and behaviors beyond the toddler's mental age are also problematic. For example, a lack of pretend play skills is a hallmark of ASDs that is assessed by most diagnostic measures; however, the ability to engage in pretend play with dolls or other objects does not typically emerge until 19–22 months (Westby, 1980). Similarly, joint attention typically develops between 9 and 18 months, making this behavior a contentious candidate for autism assessment in children with mental ages less than 12 months (Swinkels et al., 2006). The absence of communication and social skills may be due to delayed development rather than specific to autism. Finally, children with low mental age likely lack sufficient cognitive development to recognize patterns or similarities among objects and events, and therefore they may not experience the distress caused by transitions or by disruptions of habitual routines that is evident in older children with an ASD (Vig & Jedrysek, 1999).

As researchers and clinicians continue their efforts to identify ASDs in younger children, it seems likely that diagnosis will focus increasingly

on early forms of social communication and reciprocal interaction, including behaviors evident very early in development which serve those functions, such as gaze shifting, eye contact, gesture, affect sharing, joint attention, and pointing. The following is a brief description of a young child who presented precisely those concerns beginning at about the age of 12 months.

Case Report: Zachary

Zachary lived with his parents, Susan and James, and his 3-year-old brother, Sam. James worked from home as a salesman; Susan was at home full time to care for the children. Zachary's older brother had received Early Intervention services for delayed language acquisition, but he was now developing typically at the age of 39 months.

Zachary was the product of a healthy, planned, and uncomplicated pregnancy. He was born at term after a vaginal delivery and weighed 8 pounds, 10 ounces. He was bottle fed and spit up often. He continued to gag easily as a toddler. When Zachary was 4–5 months old his parents became concerned that his head was unusually large. They had him examined by a neurologist, who found no abnormalities or cause for concern. Zachary was re-evaluated by the same neurologist at 12 months old, and no abnormalities were identified. Other than those issues, Zachary was a healthy and happy infant.

Susan and James report that Zachary was a smiling and responsive baby throughout his first 6 to 8 months of life. He attained motor milestones at the expected times: he sat alone at 9 months and walked at 13 months. At about 10 months, Zachary's parents began to notice a decline in his responsiveness to social play and in his eye contact. They report that until that time, Zachary had enjoyed playing games such as patty-cake, made consistent eye contact, and was easily engaged in social play. For the first 11 months of Zachary's life, his older brother had received Speech and Language Therapy services in the family's home. The speech pathologist had frequent contact with Zachary. Neither she nor his parents noted any concerns with his social development or early communication skills.

When Zachary was between the ages of 12 and 15 months, his parents' concerns grew more serious and more focused. Zachary seemed to lose interest in social exchanges and made less frequent eye contact. He began to resist being held and began arching his back when picked up. He lost interest in toys he had previously enjoyed. At 15 months his primary play interest was in pop-up toys. Zachary babbled expressively, but he had no words and used no communicative gestures.

Zachary was nearly 16 months old when he was referred for evaluation following his failure on the Modified Checklist for Autism in Toddlers. This is earlier than the 16–30 month range recommended by the authors of the M-CHAT, but both Zachary's mother and his pediatricians had concerns with his development by 15 months. Zachary's mother spoke to his pediatrician at his 15-month well-baby visit about her concerns that Zachary was slow to develop language and appeared to be more self-absorbed and less interactive over time. The pediatrician referred the family to early intervention services, who subsequently screened Zachary for early signs of autism and referred him for further evaluation.

Zachary was evaluated in two sessions separated by a few weeks. Both of Zachary's parents were present throughout both evaluation sessions. His parents were administered the Autism Diagnostic Interview and the Vineland Adaptive Behavior Scales. Zachary was evaluated using the Mullen Scales of Early Learning and the Autism Diagnostic Observation Schedule. The evaluating clinician also completed the Childhood Autism Rating Scale and a *DSM-IV* autism symptom checklist.

Zachary's parents' responses to the Autism Diagnostic Interview provided further elaboration of their concerns about his atypical development. He communicated primarily by nonspecific crying, although he occasionally pulled his mother's hands to indicate that he wanted food. On occasion he would also pull her hand to request that she activate a favorite toy. He had no conventional gestures, nor did he point at objects outside his reach or follow a point. On occasion he would point to pictures in a book. Zachary appeared to understand very little language directed at him, with the exception of the words *no* and *bottle*. He did respond to a few highly familiar routines: he looked up expectantly if his parents approached him and said, "I'm gonna get you," and he calmed visibly when his mother

sang a familiar song. Zachary smiled at his mother occasionally in response to her smile, but this occurred with decreasing frequency; he was more likely to smile in response to television programs.

Zachary also developed several atypical behaviors between the ages of 13 and 15 months. He became very interested in texture and pattern. He began walking on his toes and occasionally banging his head, and he began flapping his hands by his side when he was excited and when he watched television.

Observation of Zachary throughout the evaluation confirmed many of his parents' concerns. With encouragement and visual prompting, Zachary was willing to attempt many of the tasks presented to him, although he did not appear interested in the tasks, and he did not persist at tasks he found difficult. His parents reported that his behavior throughout the evaluation sessions was largely typical of his presentation more generally.

Zachary's scores on the Mullen revealed considerable variability in his skills. On the Gross Motor Scale he attained a T-score of 46, which fell within normal limits for his age. He had more difficulty with the Fine Motor tasks for which he received a T-score of 33. But he had very significant delays in the areas of nonverbal skills (Visual Reception Scale), expressive language, and receptive language, where his skills all fell nearly three standard deviations below the mean. Zachary's ability to communicate intentionally and to understand language was estimated to fall at the 7–9-month level. Zachary was largely disinterested in the tasks presented by the Visual Reception Scale of the Mullen and did not appear to understand the directives. His score on this scale fell at the 6-month level.

Zachary's delays were corroborated by his parents' responses to the Vineland Adaptive Behavior Scale. Zachary's motor skills appeared to be intact, as evidenced by his Standard Score of 119 on this scale. He attained a Standard Score of 81 on the Communication Scale, consistent with an age equivalent of 10 months, and with his scores on the Mullen Language Scales. Zachary's parents described his socialization skills as consistent with those of a 9-month-old, which earned him a Standard Score of 83 on the Socialization Scale. Finally, his Daily Living Skills earned a Standard Score of 76 and an age equivalent of 8 months.

Clearly both the developmental assessment and Zachary's parents' report of his adaptive skills depicted a highly atypical developmental pattern. Zachary's motor skills appeared to be largely intact, while both his language development and his nonverbal skills were markedly delayed, along with his adaptive skills and play skills as indexed by the Vineland.

Zachary's responses to the play probes of the Autism Diagnostic Observation Schedule corroborated many of the social concerns his parents described. He was briefly interested in many of the toys available to him, but he did not engage in purposeful play with the single exception of pushing buttons on a pop-up toy. He preferred to wander around the room, fingering one toy after another and looking at the pattern in the carpet from a variety of visual angles. It was possible to engage Zachary in interaction for very brief periods of time and in highly familiar routines. For example, he was willing to play a vocal imitation game for two turns with the examiner, before he wandered away from interaction and resumed studying the carpet. Zachary did not respond to his name or to social smiles directed at him by his mother and the examiner. He did respond to his mother singing a familiar song by turning toward her and smiling. He made infrequent and very fleeting eye contact with all of the adults present. Zachary clearly enjoyed some of the toys presented during the ADOS. For example, he watched the bubbles, but he did not make eye contact, nor did he request more bubbles. Instead he wandered away from the table. Zachary had no interest in any pretend play activities, and he did not make any communicative overtures during the course of the ADOS. He did not attempt to call attention to his activity, to direct adult attention to interesting objects, or to share his pleasure or excitement. Zachary received a Total Communication and Social Interaction Score on the ADOS of 20, well above the cutoff for a diagnosis of Autistic Disorder.

Zachary received a score of 32.5 score on the Childhood Autism Rating Scale, which placed him in the mildly-moderately autistic range. He clearly met *DSM-IV* Criteria for a diagnosis of Autistic Disorder. He exhibited marked

impairment in reciprocal social interaction, qualitative impairments in communication, and stereotyped and repetitive motor mannerisms and persistent preoccupations with parts of objects.

Zachary's parents were clearly devastated but not surprised by his diagnosis. They also expressed relief that their growing concerns had been validated and that a definitive diagnosis might provide guidelines for effective intervention.

Within weeks of receiving a diagnosis, Zachary was enrolled in intensive early intervention services. He received 20 hours of individual intervention weekly, including weekly speech and language services, weekly occupational therapy and intensive behavioral intervention in his home. At age 2 he began attending a small mother–child play group in addition to his in-home services. At age 3 Zachary began attending a small preschool program for 23 hours weekly. He continued to receive Speech and Language Therapy as well as Occupational Therapy Services, although he no longer received individual behavioral instruction. He did receive individual instructional support designed to facilitate his interactions with peers. Zachary's parents further supplemented his program with intensive work with him at home, and with physical therapy and therapeutic horseback riding.

Zachary was re-evaluated as part of a follow-up study when he was 53 months old. He was administered the same series of measures with the exception of the Vineland Scales and a portion of the Mullen. These were not completed because Zachary became ill.

Zachary's mother completed the Autism Diagnostic Interview and described marked progress in his social and communicative skills. At the age of 24 months Zachary began speaking single words and his language has progressed to full sentences, question forms, and reciprocal conversation. Occasionally Zachary repeated questions directed to him; this was most likely to occur when he was confused by the question. At times Zachary also repeated greetings when people did not respond to him initially.

Zachary's social skills improved dramatically. He made consistent eye contact without prompting and smiled readily in response to smiles. Zachary reportedly played with peers individually and in small groups and initiated play by asking children to join an activity. He was reportedly sad when children left the playground and said good-bye spontaneously. Zachary enjoyed playing with trains, but he could be redirected to other activities. He also enjoyed playing on playground equipment and reading books. He had a preferred friend at school who occasionally visited his home. Zachary engaged somewhat reluctantly in pretend play, most often when initiated by others.

Zachary no longer exhibited stereotypic movements or self-injurious behavior. He continued to struggle with a strong gag reflex and with sensory irregularities. For example, he sometimes sought spinning chairs and he actively resisted having his hair dried.

Zachary's responses to the play probes of the ADOS, Module 2, largely corroborated his mother's description of him. He made consistent eye contact, responded readily to his name, and used language to request desired activities. He showed several objects to his mother and directed her attention to objects he found interesting. He was quick to share his pleasure in preferred activities and to solicit his mother's participation as well as that of the examiners. He willingly engaged in brief periods of reciprocal conversation with the examiners and was able to describe a story depicted in a book. His descriptions were somewhat limited, and while he was able to elaborate conversation following his partners' lead, his responses were more limited than expected of a child of his age. Zachary engaged in brief periods of pretend play. He was most comfortable doing this with adults and did not enjoy play with a series of small figures. Zachary received a score of 3 on both the Communication and the Social Interaction Scales of the ADOS, resulting in a Total score of 6, below the Autism Spectrum Cut-off of 8, but still indicating some residual autistic symptoms.

Taken together, the data from both the ADI and the ADOS suggest that Zachary no longer met criteria for a diagnosis on the autism spectrum. He received a score of 22 on the CARS, which falls in the nonautistic range. He continued to struggle with sensory irregularities and with behavioral regulation, and he may have had relatively subtle language difficulties, but his social development appeared to be progressing on a more typical developmental path. He was

inattentive throughout much of the evaluation and occasionally noncompliant, and those behaviors interfered with his performance to a moderate degree.

Zachary demonstrated broadly age-appropriate skills on the Expressive and Receptive Language Scales of the Mullen. He attained T-scores of 45 on the Receptive Language Scale and 44 on the Expressive Language Scale. He communicated in four-word phrases; used pronouns; followed a series of commands; and identified numbers, letters, and number concepts. The Mullen offers a limited assessment, however, of higher order receptive language skills, and some of Zachary's behavior suggested that he should receive further evaluation in this area. Zachary attained a T-score of 35 on the Fine Motor Scale of the Mullen, but he was clearly fatigued and refused to attempt several tasks presented to him. Test administration was suspended at this point and neither the Gross Motor Scale nor the Visual Reception Scale was administered.

Zachary's case, while incomplete, illustrates many of the issues associated with the diagnosis of an ASD in young children. He was identified very early in development and presented readily apparent delays in communication skills and reciprocal social interaction. While his delays were consistent across multiple developmental areas, his age-appropriate scores in motor development, as well as his atypical social presentation, suggest that developmental delay was insufficient to explain his difficulties. His presentation was characterized by a high degree of consistency across measures and across parental report and observation. As a result, a diagnosis of an ASD could be made with a high degree of confidence.

Zachary's difficulties followed a period of apparently typical development and either developed spontaneously between 10 and 15 months, or more likely, emerged as Zachary failed to keep pace with social and communicative milestones. This regressive pattern of emergence of autism is quite common (estimates vary from 15% to 47%; Stefanatos, 2008). Zachary's developmental discontinuities could not be associated with any medical procedures, known risks, or traumatic circumstances. Nor were any underlying neurologic difficulties ever identified. Unlike some children diagnosed with an ASD in the second year of life, Zachary exhibited atypical behaviors and stereotypies as a toddler. These appeared to follow the appearance of social concerns, and they appeared to increase as his social withdrawal increased.

While it is not possible to predict Zachary's progress from this point, his gains thus far bode well for his continued positive trajectory. He may be at risk for attention difficulties, language concerns, or mild social/emotional difficulties, but his progress thus far has been impressive. The minority of children who lose the diagnosis of an ASD do seem to be at risk for attention, subtle difficulties with higher order language functions, and anxiety disorders (Helt et al., 2008; Kelley, Fein, & Naigles, 2006). Careful examination of Zachary's initial presentation reveals little that would have predicted his positive outcome thus far. Clearly, children with autism require, individual intervention services focused on behavioral strategies and functional communication skills beginning as early as possible. The initiation of those services when Zachary was 16 months old, their continuation into the preschool years, and Zachary's family's active involvement in his treatment undoubtedly facilitated his progress, although none of those factors guarantee positive outcomes.

Zachary's case underscores the critical importance of early identification and aggressive early intervention. At the same it reminds us of how much there is yet to learn about the presentation of an ASD in toddlers, the pathways the disorder is likely to take, and those factors that may mediate outcome for young children.

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4

Autism Spectrum Disorders: A Case of Siblings

Stephen M. Kanne and Janet E. Farmer

Autism spectrum disorders (ASDs) are characterized by deficits in three core areas: communication skills, social ability, and atypical behaviors. They are considered to be neurologic disorders with a complex genetic origin. Though ASDs share common areas of impairment, they represent a phenotypically heterogeneous group or spectrum of disorders with clinical presentations that differ widely across each affected individual. ASDs vary in core symptom severity, may have other cognitive deficits or other neurological conditions, and often have comorbid emotional, behavioral, and adaptive difficulties that add to their behavioral heterogeneity. Parents and professionals often seek out a neuropsychological evaluation when an ASD is present or suspected for differential diagnosis and to guide treatment recommendations.

Although the specific etiology of autism is unknown, many studies have found evidence of a strong genetic component (Cederlund & Gillberg, 2004; Lauritsen, Pedersen, & Mortensen, 2005; Miles et al., 2005; Muhle, Trentacoste, & Rapin, 2004). If a family member has an ASD, it is not uncommon that another family member has an ASD as well. For example, Cederlund and Gillberg (2004) found that 70% of individuals diagnosed with idiopathic autism had a first- or second-degree relative who also had symptoms of an ASD. The recurrence rate in siblings of children with ASD ranges up to 8%, which is much higher than the prevalence rate in the general population (Muhle et al., 2004). To approach the numbers differently, siblings of children with ASD are 22 times more likely to have an ASD compared to siblings of children who are typically developing (Lauritsen et al., 2005).

The purpose of this paper is to present cases of two male siblings, 14 months apart in age, whose parents had significant concerns regarding an ASD for both from a very early age. These brothers were raised in the same household and experienced very similar treatments and therapies. Their cases demonstrate how two children, both diagnosed with Autistic Disorder and from nearly identical environmental contexts, can vary considerably in their initial presentation, manifestation of symptoms, symptom progression, and functional skills. First, we briefly describe the components of an autism assessment, then we discuss each of the siblings' first evaluations. A brief synopsis of the 5 years between the evaluations is then offered, followed by a description of the siblings' second evaluations. Finally, we discuss several interesting facets of the cases. Given the behavioral nature of an autism diagnosis, we intentionally focus a great deal on their presentation and behaviors during the evaluations.

Autism Evaluation

In addition to collecting a detailed history reviewing early developmental progression and past symptom presentation, clinicians must assess the core symptoms of an ASD currently presenting through direct observation and interaction with the child. When possible, clinicians

should gather information from multiple settings, such as school and home, and include an assessment of other domains that explore alternative etiologies. Cognitive and neuropsychological testing can be helpful in informing differential diagnoses and guiding treatment recommendations for medical and educational professionals (e.g., Klin, Saulnier, Tsatsanis, & Volkmar, 2005; Ozonoff, Goodlin-Jones, & Solomon, 2005).

The Case of AB: Initial Evaluation

Concerns regarding developmental delays and atypical behaviors prompted AB's initial neuropsychological evaluation at 2 years, 3 months of age. The parents noted that AB was often content to play by himself and did not seek out peers (e.g., in Sunday school class). Instead, he appeared aloof and withdrawn. Though he would initiate contact with his parents and would show them some toys, this was typically done "on his own terms," and he easily became hyperfocused on toys and objects. They also had concerns about his speech and language development. AB was only speaking single words at the time of the evaluation, and these were hard to understand. He rarely pointed or gestured and would lead his parents by the hand if he needed something. He often lined toys up, spun objects (e.g., turned toy cars over to watch the wheels), and engaged in repetitive hand motions near his face.

Brief Medical and Developmental History

AB was born weighing 7 lb, 7 ounces after a full-term pregnancy with no prenatal or perinatal complications reported. As noted above, he experienced significant speech and language delays. Motor developmental milestones were achieved within normal limits; he rolled over at 4 months, sat alone at 6 months, crawled at 8 months, and walked at 10 months. He reportedly had three "fainting spells" wherein he lost consciousness after a period of intense crying. During the second episode, he appeared to stop breathing for a brief (15-second) period and his lips turned blue. An electroencephalogram (EEG) at the time was normal.

Behavioral Observations

When greeted in the waiting room, AB did not look at the examiner or return the greeting. He had some difficulty transitioning to the interview as he continued playing with toys. During the interview with his parents, AB played alone quietly, not acknowledging the examiner's presence. He did not use meaningful speech during the interview.

AB willingly left the interview room to begin testing, but he required physical prompting. As soon as the door to the testing room closed, he began to fuss and required a brief visit from his mother until he became interested in playing with foam shapes. He held the shapes up one at a time, telling the examiner the name of each shape (e.g., triangle, square, circle). After a few minutes, he was introduced to pictures to match with a scene. Instead of completing the task or attending to the examiner's instructions, he rotated the pictures, then stood up and left the testing room. After several failed attempts to re-engage, the examiner accompanied AB to the waiting room where he played with several toys but did not interact with the examiner. He eventually climbed into the examiner's lap without hesitation and fell asleep.

During the evaluation, AB was observed to flap his hands while standing still and when running. Occasionally, he ran on tiptoes and with his toes slightly turned inward. He also postured his hands in an odd manner. He spoke very little throughout the evaluation. Whereas some of his words were difficult to understand, others were quite clear. He clearly enunciated the names of shapes and counted in English, Spanish, and German. He rarely responded when the examiner called his name. In fact, he appeared to be unaware of the examiner except when toys were mentioned or when he wanted something. At these times, he took the examiner's hand and led to what he wanted.

Evaluation Results

AB was unable to engage in the formal cognitive testing. Given his speech and language delays, the Leiter International Performance Scale, Revised (Leiter-R; Roid & Miller, 1997) was attempted but then discontinued due to his lack

of engagement. His results on the Autism Diagnostic Observation Schedule (ADOS; Lord, Rutter, DiLavore, & Risi, 2002) met the criteria for Autism (Module 1, Raw = 19). The ADOS places the child in several situations, or "presses," designed to elicit social and communicative responses allowing the examiner to assess core autism symptoms.

AB's overall adaptive skills were in the impaired range using the Vineland Adaptive Behavior Scale (Sparrow, Cicchetti, & Balla, 2005) by parent report (Adaptive Behavior Composite = 64). Parents also noted clinically significant internalizing behaviors on the Childhood Behavior Checklist (CBCL; Achenbach & Rescorla, 2001). The Childhood Autism Rating Scale (CARS; Tobing & Glenwick, 2002) was completed to better understand AB's history and developmental progression. AB's results were in the mild–moderate autism range (Total Score = 30.0).

As a result of the evaluation, the clinician determined AB met diagnostic criteria for the presence of autistic disorder of at least moderate severity. The parents participated in a comprehensive feedback that explained the results, diagnosis, and answered general questions regarding ASD. Recommendations included a suggestion that AB undergo a full speech/language evaluation with subsequent therapy and receive an evaluation to secure special education services. In addition, the examiner encouraged the family to pursue applied behavior analysis (ABA) therapy for AB, an intense behavioral program proven efficacious for children with ASD (c.f., Myers, Johnson, & Council on Children with Disabilities, 2007). Finally, the parents received information about multiple resources, including contact information for parent support groups and educational resources such as books and Web sites.

The Case of CD: Initial Evaluation

AB's older brother, CD (3 years, 5 months), was evaluated the day after AB. Similar to his brother, CD had several areas of concern. Parents reported that CD was aloof around other children, rarely initiated interactions, and was often withdrawn. He did not engage in parallel play, but he was content to play alone. He was described as affectionate and "connected" to his parents, but he typically had difficulty demonstrating empathy. His eye contact was poor, and he had trouble coordinating his gaze appropriately with his vocalizations. His parents indicated their perception that CD was more "severe" than AB, as he showed similar social difficulties but was more reactive with problematic behaviors.

CD had a history of developmental speech and language delays. Although he was able to speak single words and phrases in a normal developmental time frame, he did not use language for communicative intent. Instead, he used language only to label things or in a rote manner. He engaged in frequent echolalia (i.e., repeating words or phrases he has heard) and did not engage in reciprocal conversation. His parents reported that his nonverbal communication was also poor. He was not gesturing in a typical manner or using facial expressions effectively.

At the time of the evaluation, CD's parents reported that he sometimes banged his head, but not often. However, his play was described as repetitive and mechanical. He tended to be interested in only one main topic at a time. When playing with toys, he often engaged in a repetitive ritual (e.g., turning it in a certain sequence), and he often lined up and categorized his toys. He relied heavily on structure and routines, and they noted he often became upset when making transitions. He had some mild tactile defensiveness (e.g., to textures and clothes, dislikes getting a haircut or feeling the clippings on his skin). He insisted on having his hands cleaned if he perceived them to be dirty.

Brief Medical and Developmental History

CD was born weighing 7 lb, 0 ounces after a full-term and uncomplicated pregnancy. His early motor developmental milestones were reached in a timely manner: rolling over at 4 months, sitting alone at 5 months, crawling at 8 months, and walking at 10 months. In contrast, his parents noted delays in CD's early speech and language developmental milestones. Although he spoke single words at 7 months, his progress from that point forward was described as "slow." They noted that at age 3, he was not speaking in sentences or using speech to communicate in a

meaningful way (e.g., he was primarily naming objects).

Two months prior to the evaluation, CD had participated in an evaluation within his school district, wherein his nonverbal intellectual functioning was in the low average range (Leiter-R; standard score [SS] = 85). He performed in the impaired range (SS = 65) on every aspect of the Battelle Developmental Inventory (Newborg et al., 1988), with the exception of the specific subdomain Coping (SS = 100). His preacademic skills were in the superior range (Bracken Basic Concept Scale–Revised; Bracken, 1984). He had an Individualized Educational Program (IEP) and was receiving speech/language therapy with goals addressing his social skills.

Behavioral Observations

When CD was greeted in the waiting room, he made fleeting eye contact but did not maintain it and did not return the greeting. During the interview with his parents, CD explored the room actively and played by himself. He did not acknowledge the examiner's presence or reference others in the room. He spoke using single words and some phrases, though many of his phrases were echolalic. He required prompting to transition to the testing room.

When CD began the testing, he repeated "That silly [examiner's name]" over and over to his father. Upon entering the waiting room during a break in testing, he threw himself to the floor and began crying. His father was able to calm him, after which he accompanied his father back to the testing room. After a brief period with CD on his lap, his father attempted to leave, at which time CD reached for him and began to cry again. Shortly after his father left the room, CD climbed into the examiner's lap and resumed testing. As testing progressed he stopped speaking and crawled to the floor. He was taken back to the waiting room where he had the same reaction as earlier.

Throughout the evaluation, CD rarely produced reciprocal language. He named many things, but primarily echoed the examiner's statements and questions. His many sound substitutions made him difficult to understand. When he appeared to know the answer to a question, he answered quickly. At other times, he looked away from the test material, fell onto the floor, and squirmed. At these times, he did not respond when the examiner asked if he knew the answer. He also did not seem affected when the examiner told him it was "okay" to tell if he did not know an answer. Similarly, he did not show a response to positive statements made by the examiner. Though often difficult to engage, overall, CD was able to be redirected and to complete the testing.

Other notable behaviors included calling his parents by their first names, which they reported was typical. During breaks, he did not interact with the examiner until the end of the day, during a game of imaginative play initiated and maintained by the examiner.

Evaluation Results

CD's overall level of intellectual functioning was in the average range (SS = 103) (Differential Abilities Scales; Elliot, 1990). His performance on the subtests ranged from low average to high average: Block Building SS = 94; Verbal Comprehension SS = 85; Picture Similarities SS = 115; Naming Vocabulary SS = 117. He was also administered a measure of basic language skills, the Clinical Evaluation of Language Fundamentals–Preschool (Wiig, Secord, & Semel, 1992). His receptive language was in the low average range (SS = 81), and his expressive language was in the low end of the average range (SS = 90). As Table 4-1 demonstrates, his individual subtest scores were variable.

Table 4-1. CD's Speech/Language Results on the Clinical Evaluation of Language Fundamentals–Preschool

Index Scores	Standard Scores
Receptive Language Score	81
Expressive Language Score	90
Total Language Score	85
Subtests	**Scaled Scores**
Linguistic Concepts	5
Basic Concepts	10
Sentence Structure	4
Recalling Sentences in Context	8
Formulating Labels	10
Word Structure	7

Parents reported CD's overall adaptive skills to be in the borderline range on the Vineland Adaptive Behaviour Scale (Adaptive Behaviour Composite = 79) and noted clinically significant internalizing behaviors (CBCL). Teachers also reported a significant degree of internalizing behaviors (Teacher Report Form). Report of his developmental progression met the criteria for mild to moderate autism (CARS; Total Score = 32.5). Of note, on the Gilliam Autism Rating Scale (Gilliam, 1995), his parents did not endorse a significant amount of difficulties, resulting in a "low probability" of autism on this measure. CD's results on the ADOS met criteria for autism (Module 2; Raw = 21).

Similar to his brother, evaluation results indicated that CD met diagnostic criteria for the presence of autistic disorder. Cognitive testing ruled out an overall developmental delay and mental retardation. Results also suggested that a speech/language disorder alone did not account for his presentation. Though his basic language skills appeared relatively intact, his functional and pragmatic language skills were significantly deficient. Recommendations were nearly identical to those for his brother, but with additional suggestions for managing his reactivity.

Comparison of Siblings at Initial Visit

AB and CD both had clear deficits in the three areas necessary for a diagnosis of autistic disorder, reflected in their formal results (e.g., ADOS, CARS); however, they presented in vastly different ways. Table 4-2 presents a comparison of the siblings' results after the initial evaluation. Both boys had significant speech/language delays, but CD's language was better developed, although it was primarily nonfunctional and echolalic. Both boys had significant social deficits, although CD was able to engage better, and even completed testing; his overall response style was negative and reactive. Both boys also had significant atypical behaviors, though AB demonstrated many more repetitive behaviors, including hand flapping, tip-toe walking, and atypical hand movements, whereas CD adhered more to routines and demonstrated stereotyped language. Both children had difficulty with eye contact; however, AB tended to avoid eye contact, whereas CD had more difficulty coordinating his gaze with his vocalizations. Whereas the parents felt that the older boy, CD, was more severely affected due to the extent of his behavioral difficulties, our results suggested that the younger child was more "severe" due to his degree of aloofness, his inability to engage, and the nature of his repetitive behaviors.

Interim

The parents brought both brothers back for a re-evaluation 5 years after the initial evaluation. In the interim, the parents had moved geographically, actively sought information regarding autism, and proactively involved both brothers in many different types of therapy. In addition to having both children participate in ABA therapy and speech/language therapy, the family also pursued many alternative and biomedical treatments, including a gluten/casein-free diet, vitamin supplements, chelation, and the use of a hyperbolic oxygen chamber. They reported significant improvements for both boys over time.

The Case of AB: Second Evaluation

At the time of the second evaluation, AB was in kindergarten, and 7 years, 3 months of age. He had reportedly made marked improvements in almost every area of concern over the intervening five years. He was receiving speech/language,

Table 4-2. Comparison of Siblings after First Evaluation

	AB	CD
Intellectual Functioning (Leiter-R)	Unable to engage	Average (SS = 103)
Adaptive (Vineland)	Impaired (SS = 64)	Borderline (SS = 79)
Autism Diagnostic Observation Schedule	Total Score = 19	Total Score = 21
Childhood Autism Rating Scale	Total Score = 30	Total Score = 32.5
Speech/Language (CELF-P)	Unable to engage	Low Average (SS = 85)

occupational therapy, and physical therapy as part of his IEP under the special educational eligibility category of Autism. His parents noted that his academic skills were well developed. Both his verbal and nonverbal communication skills were greatly improved, and he now spontaneously initiated conversation with others and responded reciprocally. Though he typically conversed in areas of his special interest, or introduced a conversation with a script specific to a person, he had begun to ask appropriate questions during conversations. Many of his more obvious atypical behaviors had decreased in frequency or intensity.

However, his parents remained concerned regarding AB's ability to communicate effectively. He engaged in echolalia when not fully following a conversation, and his scripting interfered with many interactions. When upset or emotionally charged (even happy), atypical behaviors returned, such as hand flapping and spinning. He hit his head with his fist when he got upset, and, on his "bad days," could be very rigid and reactive. He continued to become self-absorbed and hyperfocused, and he often needed prompting.

Several "splinter skills," or areas of ability well above his other skills had emerged. Some of these were nonfunctional, such as naming all the Presidents of the United States with supporting details. He also had demonstrated a high degree of musical talent. In fact, the parents brought a roll-out keyboard that AB played (with seeming proficiency), though he never had music lessons, and a recording of songs he created using a software program wherein he played each separate instrument himself.

Behavioral Observations

When greeted in the waiting room, AB made eye contact and returned the greeting, but he spoke in a scripted manner with formal intonations. He engaged in some reciprocal conversation, but he did not coordinate his gaze and quickly changed the topic to Presidents of the United States. He demonstrated his ability to recall any President by number.

AB's prosody was exaggerated and had a musical or sing-song quality, particularly when repeating fairly well-rehearsed social responses. He had an extensive vocabulary and mimicked phrases learned from television in stereotyped phrases (e.g., "get out of town… check out our resorts"). He also occasionally made comments that were out of context or possibly paraphasic (e.g. "that looks like a rain job" when referring to an image resembling a planet). He produced sound effects, such as crash sounds, or repeated utterances, such as "tooka took, tooka took, tooka took" or "whoota, whoota, whoota" multiple times during the evaluation. If a question was asked of him, he answered appropriately and spontaneously offered elaborations; however, he rarely asked questions of the examiner.

During testing, AB was able to engage and was pleasant and cooperative. He often used verbal mediation (i.e., talking his way through a problem) during visual tasks. He scanned visual stimuli very carefully, particularly when they were detailed, and was noted to perform quite well on visual-spatial reasoning tasks. However, on items that were timed, he became tense and responded in a more haphazard manner, focusing more on completing the items quickly rather than correctly. AB often created drawings out of words or numbers that he wrote. For example, after spelling the word *look*, he drew a pair of glasses out of the two "Os." After he thought of different types of "silly" high-fives, he suddenly stopped and said "ok, let's concentrate." He also referenced the examiner's facial expressions on multiple occasions. He supported his verbal communication with nonverbal gestures, such as pointing and shrugging. AB exhibited several repetitive behaviors during the evaluation, such as hand flapping and staring at himself in the one-way mirror.

Evaluation Results

AB had many areas of marked improvement compared to his results from 5 years prior. He was able to engage in formal testing, and his overall level of nonverbal intellectual functioning was in the superior range (Leiter-R; SS = 127). His academic skills were also strong (WIAT-II; Wechsler, 2002), with word recognition and spelling in the very superior range, and computational skills in the average range (e.g., Reading SS = 138, Spelling SS = 153, Numerical Operations SS = 103). His visual motor integration was in the average range (SS = 103) (The Beery-Buktenica Developmental Test of Visual-Motor Integration,

Table 4-3. Comparing AB's Diagnostic Algorithm Raw Scores to Current Algorithm Raw Scores on Each ADI-R Subscale

ADI-R Subscale	Diagnostic Algorithm	Current Behavior Algorithm
Reciprocal Social Interaction (Cut-off = 10)	20	7
Communication (Cut-off = 8)	21	10
Repetitive Behaviors and Stereotyped Patterns (Cut-off = 3)	10	8

5th Edition [VMI]; Beery, Buktenica, & Beery, 2004).

A structured interview of past and current autism symptoms, the Autism Diagnostic Interview, Revised (ADI-R; Lord, Rutter, & Le Couteur, 1994), reflected improvements in AB's autism related features. In each domain, as Table 4-3 demonstrates, his current scores indicated improvements compared to his past scores.

On the ADOS, AB's results were now in the range of an Autism Spectrum Disorder rather than meeting criteria for the more severe Autism (Module 3, Raw = 7). Thus, assessment of his current symptoms, in clinic and by parent report, indicated significant improvements in his ASD-related symptoms. However, despite these improvements, his parents' report of his adaptive skills (ABAS-II; Harrison & Oakland, 2003) remained in the impaired range (SS = 69).

AB's diagnosis remained Autistic Disorder. Recommendations targeted improving his pragmatic language and social skills, while continuing to support his need for services in school. Those involved in his care were encouraged to find ways to take advantage of his cognitive strengths and other areas of highly developed skills. Parents were also complimented for their strong advocacy and support.

The Case of CD: Second Evaluation

CD, now 8 years, 5 months of age, had also reportedly made a great deal of progress. His parents noted marked improvements in his communicative/interactive skills, his social skills (e.g., social awareness and perception, ability to understand others' perspective, sense of humor), and his overall mood regulation. They continued to have concerns regarding his tendency to perseverate, his adaptive skills, and his emotional reactivity.

Behavioral Observations

When CD was greeted in the waiting room, he made brief eye contact, smiled, and returned to his play. While transitioning to the interview room, he engaged in casual and reciprocal conversation with appropriate coordination of eye gaze with the examiner. Verbally, he demonstrated slower response latency to questions and his verbal pacing was mildly atypical. During testing, CD was pleasant and cooperative, though he tended to be verbose and tangential. For example, when asked what a number is, he replied "It's a thing that a caveman invented to keep track of things. They were tired of saying, I have a bunch of camels… it can be a tally mark… a symbol, it can be anything, for example, two horses, five golden rings… so we don't have to carry slates and write tally marks." When asked to define an "alligator," he described various types of reptiles and named prehistoric dinosaurs, before providing the correct response. His definition of objects and concepts tended to be functional in nature, as he often provided their various uses, rather than providing a more abstract definition. CD had some difficulty on a measure of visual-spatial construction, in which he was to assemble blocks in a manner similar to that of visual model. He was aware that he was being timed for this task and appeared pressured. For a number of items, he stated that he was finished and then continued to work identifying that something was not quite correct. However, he was usually unable to figure out how to amend his design. On a test of written calculations, CD exhibited significant frustration. He sighed multiple times throughout this task, and on items that were difficult for him, he erased his answers repeatedly before committing to a final answer. It was noted that CD typically did not smile when making a humorous comment, until he saw the examiner smile or laugh, at which point he returned the smile/laugh.

Evaluation Results

On this evaluation, CD demonstrated a significant discrepancy between his level of verbal intellectual functioning, in the superior range (VIQ = 129) (Wechsler Abbreviated Scale of Intelligence; Wechsler, 1999), and his level of visual spatial intellectual functioning, in the average range (PIQ = 97). His academic skills followed the same pattern of stronger performance on verbally related tasks: word recognition in the superior range (WIAT-II; Reading SS = 126), spelling in the very superior range (Spelling SS = 137), and computational skills in the average range (Numerical Operations SS = 97). His ability to learn and recall verbal information (CVLT-C; Delis, Kramer, Kaplan, & Ober, 1994) was in the average range immediately (List A Total SS = 105) and after a delay (e.g., Long Delay Free Recall SS = 93). His visual motor integration was in the superior range (VMI; SS = 122).

Similar to his brother, CD's results on measures specific to autism symptoms reflected improvements. On the ADOS, his total score was now in the range of an autism spectrum disorder rather than the more severe Autism (Module 3; Raw = 7), and, as shown in Table 4-4, his ADI-R results also reflected improvements from the past to his current presentation.

As a result of the evaluation, CD's diagnosis was changed from Autistic Disorder to Pervasive Developmental Disorder–Not Otherwise Specified (PDD-NOS). He did not fully meet the criteria for Autistic Disorder based on his presentation at the time of the second evaluation. We felt that the diagnosis of PDD-NOS accurately captured his continued, though subtle, difficulties associated with his history of an ASD that now did not meet full criteria, while also conveying the progress that he had made. Similar to his brother, despite his improvements, CD's adaptive skills remained problematic and in contrast to his other skills: in the high end of the borderline range by parent report (ABAS-II; SS = 79).

Table 4-4. Comparing CD's Diagnostic Algorithm Raw Scores to Current Algorithm Raw Scores on Each ADI-R Subscale

ADI-R Subscale	Diagnostic Algorithm	Current Behavior Algorithm
Reciprocal Social Interaction (Cut-off = 10)	24	4
Communication (Cut-off = 8)	21	3
Repetitive Behaviors and Stereotyped Patterns (Cut-off = 3)	4	2

Comparison of Siblings at Second Visit

Both AB and CD had made considerable progress over the intervening 5 years. Table 4-5 presents a comparison of the siblings' results after the second evaluation. AB progressed from being unable to engage in testing to performing in the superior range or above on measures of academic and intellectual functioning. AB's language had improved and he was now using fluent and complex sentences, but he remained echolalic and scripted. He continued to engage in repetitive behaviors, though they had decreased in frequency and intensity.

Table 4-5. Comparison of Siblings after Second Evaluation

	AB	CD
Intellectual Functioning	Superior (Leiter-R; SS = 127)	Superior (WASI; SS = 129)
Adaptive (ABAS-II)	Impaired (SS = 69)	Borderline (SS = 79)
Autism Diagnostic Observation Schedule	Module 3, raw = 7	Module 3, raw = 7
ADI-R; Reciprocal Social Interaction	raw = 7	raw = 4
ADI-R; Communication	raw = 10	raw = 3
ADI-R; Repetitive Behaviors	raw = 8	raw = 2
Reading (WIAT-II)	Very Superior (SS = 138)	Superior (SS = 126)
Spelling (WIAT-II)	Very Superior (SS = 153)	Very Superior (SS = 137)
Numerical Operations (WIAT-II)	Average (SS = 103)	Average (SS = 97)

CD's intellectual functioning results improved from the average range to the high average range and he also demonstrated academic skills well above average. CD's language was now reciprocal though verbose, and he was coordinating the nonverbal aspects of communication better. CD remained reactive, though this was also improved.

Looking solely at their test results fails to convey the striking difference between the siblings from a clinical and behavioral perspective. Both had significant cognitive and academic strengths, and both received the same score on the same module of the ADOS. However, the two boys presented very differently. AB presented with many more atypical behaviors strongly suggesting an ASD. CD's presentation was such that, without knowledge about his prior history or a detailed understanding of his presentation, a diagnosis of an ASD may not appear appropriate. The pattern of differences between the boys at their initial evaluation was maintained, though it increased in magnitude.

Discussion

The evaluations of these siblings utilized the tools appropriate to an ASD evaluation. Core areas associated with an ASD were assessed each time, using instruments such as the ADOS and clinical observation. Developmental history and ASD symptom progression were assessed using the CARS (first visit) and ADI-R (second visit). Several other measures were utilized depending on their age and level of engagement, assessing language (e.g., CELF-P), intellectual ability (e.g., Leiter-R, WASI, DAS), memory (CVLT-C), and adaptive skills (e.g., Vineland, ABAS-II). In addition to supporting diagnostic decisions, these measures provided a framework to guide recommendations for family and educators.

In general, the boys' initial symptom patterns were maintained over the intervening years, though the degree of difference between them had increased. CD demonstrated better levels of engagement and basic social skills, better developed language, and less overt repetitive behaviors compared to his brother. Both improved dramatically between assessments, to the degree that the older brother's symptoms associated with ASD were very subtle. Both had received a great deal of intense therapy, and both demonstrated many areas of cognitive/academic strength. For both, the symptoms associated with ASD continued to impact their everyday living skills despite the improvements they had made.

The genetics of ASD suggest that there are many parents who have two or more children affected by ASD. The current cases underscore how differently two siblings can present and progress, despite having the same initial diagnosis. This was relevant in the siblings, as the parents initially thought that CD was more severe because of his more externalizing presentation, whereas they perceived AB to be less severe and "low maintenance." Our results suggested that the very factors that gave the impression of lessened severity were actually more impairing and suggestive of a more severe presentation of an ASD, which the later test results confirmed. Research providing a better understanding of autism phenotypes would help in these situations that could prompt better prognosis and treatment planning.

These cases demonstrate the limitations clinicians currently encounter with respect to ASD. The siblings made considerable progress, which was reflected to some extent in the results (ADOS and ADI-R). However, these tools had difficulty capturing the nature and nuances of their improvements. This is understandable as these tools were designed for diagnostic purposes and not designed to be sensitive to change. This is reflected in results wherein both boys scored similarly on the ADOS during the second evaluation, but they were immensely different with respect to their symptom presentation. Another problem is associated with both boys' very strong cognitive test results. Strong cognitive skills are associated with positive prognosis, but they may also cause others to overestimate the extent of difficulty associated with their ASD symptoms. Despite their strong cognitive skills, AB and CD struggle with day-to-day tasks.

AB and CD's cases also underscore how much remains unknown regarding ASD and how clinicians are unable to predict the outcome for children. In the current state of the field, evaluations lead to diagnosis and recommendations, but these recommendations are necessarily general and not finely tuned to each child. Identifying phenotypes can lead to specific treatments

tailored for a specific type of ASD. Without this level of specificity, many parents do as AB and CD's parents have done: pursuing multiple treatments and expending a great deal of resources not knowing which is working or why. Future research will also help determine whether their improvements were the result of their natural developmental progression, the therapies they participated in, or some combination of the two.

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5

Dyslexia in a Young Adult

Robert L. Mapou

Dyslexia is a developmental reading disorder characterized by difficulty sounding out and reading single words fluently. It is the only developmental learning disability for which there is a research-based definition (Fletcher, Lyon, Fuchs, & Barnes, 2007). This definition is as follows:

> Dyslexia is a specific learning disability that is neurobiological in origin. It is characterized by difficulties with accurate and/or fluent word recognition and by poor spelling and decoding abilities. These difficulties typically result from a deficit in the phonological component of language that is often unexpected in relation to other cognitive abilities and the provision of effective classroom instruction. Secondary consequences may include problems in reading comprehension and reduced reading experience that can impede the growth of vocabulary and background knowledge. (Lyon, Shaywitz, & Shaywitz, 2003, p. 1)

Because of early intervention or the development of compensatory strategies, a large proportion of adults with dyslexia can develop average or better ability to decode and read single words in isolation, especially if speed is not stressed. Nonetheless, adults with dyslexia typically struggle with fluency and comprehension when reading text. Even if they can read single words in isolation, they remain slow and inefficient readers. They may also have difficulty on timed measures of decoding and single-word reading. Consequently, assessment must include timed measures of reading (S. Shaywitz, 2003; S. E. Shaywitz & Shaywitz, 2005). Adults with dyslexia can also have associated difficulties in a range of language skills, including word retrieval, naming speed, verbal working memory, vocabulary, listening comprehension, and semantic knowledge (Mapou, 2009). Moreover, these skills may be more predictive of poor reading in adults than phonological awareness, which appears to be more important for acquisition of reading in early schooling.

Clinically, Wasserstein and Denckla (2009) have proposed three types of reading disorders in adults. The first is a pure phonological subtype, in which reading aloud is impaired, but comprehension is stronger. A case example of a physician with this type of reading disorder was recently presented by the author (See Appendix in Mapou, 2009). The second is a comprehension subtype with the opposite profile: comprehension is impaired, but reading aloud is stronger. The third is a combined subtype, in which both reading aloud and comprehension are impaired. In the case presented here, a young adult college student, comes closest to this third subtype, although his comprehension was still stronger than his decoding and single-word reading skills.

Case Study: Mr. C

Mr. C, a 19-year-old community college freshman working toward an associate's degree, was referred by the disability support service (DSS) coordinator at the college he was attending. Historically, he had developed motor skills normally but had been slow to develop speech and language. He was subsequently slow when learning to recite the alphabet. Because his father had