CHRISTINE EVANS
FOREWORD BY PETER HARPER

Genetic Counselling

A PSYCHOLOGICAL APPROACH

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Genetic Counselling

A Psychological Conversation

The role of a genetic counsellor is to mediate between the rapid advances in molecular medicine and an individual's ability to understand and manage the risks of their inheritance. Counsellors therefore, need to be fully in command of the psychological impact of their communications. Written by a psychiatrist who later became a psychotherapist, this book is essential reading for counsellors of all disciplines. It examines the psychological processes involved and uses the framework of attachment theory to explain why people approach and respond to genetic counselling differently. Effective counselling requires a knowledge of the principles from individual and family therapy. In particular an in-depth understanding of empathy enables the counsellor to help the individual contain anxiety and process grief, and so facilitate decision-making or help with the effects of having a test result. The effect of counselling on the counsellor is examined creatively in order to enrich the interview with clients and the concept of non-directiveness is discussed in the context of similar approaches in family therapy and psychoanalysis. The theme of the professional relationship and its importance is seen as the most important factor.

Christine Evans trained as a psychiatrist and worked with disturbed adolescents before retraining as a psychotherapist. She has 10 years' experience of working with genetic counsellors. Her work includes experience as a Child and Adolescent Psychiatrist and as a Psychotherapist. Her particular interest is the integration of the psychological and physical aspects of medicine. Her present practice involves working in individual psychotherapy and teaching and running workshops on attachment theory and its use in consultations in medicine, psychiatry and psychotherapy.

Genetic Counselling

A Psychological Conversation

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Time past and time present
Are both perhaps present in time future,
And time future contained in time past.
If all time is eternally present
All time is unredeemable.

T. S. Eliot. *Burnt Norton*

To Dan and our genetic future
Richard, Hywel and Jo;
Emily and Lowri

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Acknowledgements

This book began when Helen Hughes introduced me to the world of clinical genetics at The Institute of Medical Genetics, Cardiff. The actual production has taken a long time, crystallising the ideas and putting the work on paper. It presents my understanding of the clinical work very many people shared with me. They are too numerous to name individually, but I thank everyone who took part in the teaching and supervision sessions we organised and hope the book reflects their commitment to patient care. I have tried to protect the confidentiality of patients and our discussions by building in an element of fiction.

Some people have been particularly helpful. Peter Harper, who not only supported my work with the supervision group, but also seeded the idea of recording it and contributed by writing a foreword. Jonathon Gray and Annie Procter have provided ongoing encouragement and support at times of my failing spirit and Helen Hughes has generously given time to provide comments, phraseology and advice.

My psychological friends and colleagues have provided suggestions, support and encouragement, and tolerated my preoccupation with this project. In particular, Eddy Street, who has helped on many occasions with the macro-editing to help me find a meaningful structure to present the ideas. My thanks to my friends, Steve Bowkett, who has worked with me to improve phrase and construction and Helen Rowlands, who has the ability to detect missing links of thought. I have particularly valued the conversations with Mark Rivett, Srikant Sarangi and Jeremy Holmes. Some friends have not performed any particular role but helped sustain me.

No book is written without a considerable degree of time, hard labour and self-absorption which has taken me away from family and friends. My thanks to all but, in particular, Dan, who generously provided a safe place for me to explore my own thoughts and uncomplainingly supported me by his involvement in the many difficult tasks such as referencing accurately as well as generally keeping the show on the road.

Foreword

Conversation and communication lie at the heart of genetic counselling. How well we are able to communicate will to a large extent determine how successful we are in providing answers to the questions and problems posed by those whom we see in genetic counselling practice. We hope and believe that we are already skilful in the processes involved but, at the same time, most of us recognise that we could do much better and can think of many occasions when we have been left with a sense of inadequacy, or even failure.

This book will be of great support to all involved in genetic counselling, giving both general and specific information that relates directly to its practice. The general themes explored will be especially helpful to those many people whose training has involved little theoretical knowledge of psychology and psychotherapy. By exploring this general background, notably attachment theory, in the specific context of genetic counselling, Christine Evans introduces the reader gently to the concepts involved. As a result, one comes to understand the fuller significance of factors that most of us have already intuitively recognised, but have not been able to name or relate to general principles. As she states in her preface, 'It is a way of giving back to the world of genetics what actually belongs to it'.

At a more specific level, the many quotations from individual consultations show how often the counsellor can adapt an interview to make it more fruitful, even therapeutic, using simple ways to remove blocks or open up important paths. Encouragingly, the message is that those of us in genetic counselling are often doing this already, though unconsciously; greater awareness of these approaches will allow us to use them more systematically and more confidently.

For many years I have had the great privilege of having the author as a colleague, and of being able to learn from her directly. It gives me the greatest pleasure that by writing this book she has made her experience and wisdom available to the wider genetic counselling community. Whether as professionals or as families with genetic disorders, we shall all benefit from the insights and direct help that this valuable book contains.

Peter Harper,
University Research Professor in Human Genetics,
Institute of Medical Genetics,
Cardiff University.

This book helps the counsellor understand how the personal history and emotional dynamics of the individuals who to seek help and information have the potential to inhibit the quality of communication and emotional attunement in a counselling interaction. These delicate processes are addressed by a process of critical observation, analytical description, and psychological translation of genetic counselling. The book is a valuable resource providing a deeper understanding of what is being done well and why. It is a working manual that aids two key areas of the counselling process: namely, the counsellor's knowledge and management of self, and the development of strategies that facilitate the provision of a secure and flexible framework within which the genetic counselling process can provide for those who access it.

Annie Procter
Consultant Clinical Geneticist and Clinical Director,
Institute of Medical Genetics,
University Hospital of Wales, Cardiff.

In this remarkable work, Dr. Chris Evans, a psychoanalytic psychotherapist, family therapist, and child psychiatrist, brings together her psychological expertise with long experience of working with clients and counsellors in a genetic counselling unit. The result is a book that resonates far beyond its

chosen subject. Dr. Evans shows, in her own words, how genetic counselling is more than an exchange of technical information, it is an encounter between people and about people, their pain, their fears, and their relationships. Packed with illuminating and moving clinical examples, she explores the ways in which effective counsellors create a psychological space within which listening, tracking, empathy, and metaphor can help clients make choices, come to terms with their difficult feelings, and resolve conflicts. Dr. Evans's tone throughout is wise, calm, caring, considered, and unsentimental. She has an exemplary gift for making complex psychological concepts such as defense mechanisms and attachment styles understandable and relevant to everyday clinical work. In bringing together psychology and medicine, she is pioneering the rediscovery of a lost art. This book is essential reading not just for those in the immediate field of genetic counselling, but for all practitioners who want to deepen their understanding and skills in the art of communication in medicine.

Jeremy Holmes
Consultant Psychiatrist/psychotherapist Devon NHS Partnership Trust, and Professor of Psychological Therapies, University of Exeter, and Psychoanalysis Unit, University College London.

Dr. Christine Evans has written a contemporary textbook that beautifully melds the fields of genetic counselling and psychotherapy. As a psychiatrist, Dr. Evans accurately conveys the subtleties and nuances of the struggles and decisions faced by genetic counselling clients. Her active participation in genetics cases and extensive review of transcripts offers cases to illustrate the stress and coping and attachment theories that she promotes as frames for understanding clients' reactions to genetic information. Genetic counselling graduate students and practicing genetics professionals alike will benefit greatly from this text that advances genetic counselling as the compassionate relationship it ought to be.

Barbara Bowles Biesecker
Director, JHU/NHGRI Genetic Counseling Graduate Program, and Associate Investigator, Social and Behavioral Research Branch, NIH Bethesda, Maryland.

Preface

The practice of medicine has a long tradition of making a diagnosis by building up a composite picture of the symptoms and their history, a clinical examination and specialist investigations. A diagnosis enables the course of the disease to be predicted and the appropriate treatment used. However, scientific developments in genetics, by allowing us to detect specific abnormalities in chromosomes and in individual genes, are beginning to throw light on the mechanisms involved at a biochemical and molecular level. As a result the genetic and environmental factors involved in many disorders can be separated. This has heralded the birth of the practice of clinical medical genetics, a relatively new speciality in the wide and general field of medicine. With this development not only is there is a new understanding of diseases, but also an alteration in the way medicine is practised. The availability of newly discovered genetic knowledge, to all who want it, has necessitated a shift in the dynamic between the individual patient and the professional. The days have gone when the doctor was the benevolent patriarch knowing the secrets of diseases and administering to the patient, the grateful receiver. Today, people want to take personal responsibility for their health and are encouraged to do so. There is a general interest in understanding health matters and a common desire to know about medical matters and this is validated by the individual's right of access to personal information. The individual now has choice. In particular, there is the possibility of having personal genetic knowledge of present disease, or future disease potential. In the past, most genetic tests were undertaken to determine personal risk relating to reproductive decisions. However, the identification of mutations which predispose to adult-onset disorders has increased the demand for testing and an understanding of individual risk. Medical genetics is a modern speciality in tune with today's

society, upholding individual autonomy and the right to have personal information. This is balanced by an awareness that there are consequences to accessing such information, especially as it is now possible to make a genetic diagnosis presymptomatically. The speciality has taken medicine into the future and requires a revision of the definition of the patient from 'symptom bearer' to that of 'enquirer'.

Genetic counselling has emerged to respond to the individual seeking genetic information and has taken up the challenge of how the knowledge of the genetic contribution of a disease is shared with individuals and families. This automatically places genetic counselling as an educative and communicative speciality. It is practised by professionals from different disciplines: medically trained clinical geneticists and genetic counsellors who may have come from a nursing tradition or have been more specifically trained as genetic counsellors. During the process of genetic counselling all of these professionals take the role of a counsellor irrespective of their original training. This role can be likened to a translator or a bridge-builder who carries scientific information from the laboratory into the clinical arena and makes it comprehensible and personally relevant to individuals and families.

However, the information is not given as a lecture, but as part of a dialogue between the individual and the counsellor. In the dialogue there is a two-way process with the counsellor and patient mutually influencing one another. It is a human encounter and the reciprocal interactive field is an important dynamic which needs to be included in the counsellor's analysis.

The central elements of a genetic consultation include risk assessment, information-giving, decision-making and assessment of psychological coping processes. It can take many forms depending on the nature of the disorder, the characteristics of the individual or family and the orientation of the counsellor. The complexity, variety and different ways of practising are reflected in the different definitions available. Nevertheless, within the variety of practice, genetic counselling has developed a core form and structure which includes conveying degrees of information, whilst also guiding the individual through a self-reflective process. In most consultations the discussion includes the nature of the disorder, the family history and an assessment of the individual. In keeping with this, the counsellor has developed a repertoire of skills, as an educator or informer and also as a facilitator, which in combination give the genetic counsellor a specific identity.

From the examination of transcripts, where it is possible to explore what actually happens in the many different genetic consultations, a general pattern can be discerned. However, the nature of the disorder introduces a particular emphasis and this determines the particular shape and form of the consultation.

In this book a psychological framework is presented which explains and explores the experience for the individual and the family. It clarifies how the experience is influenced by the human encounter with the counsellor. The framework provides a theoretical base and a language to enrich and inform the counsellor's practice and understanding. It invites genetic counselling to use that language to sit alongside the scientific language of genetics.

The genetic counsellor does not need to be a psychotherapist, but the practice needs ideas taken from counselling and psychotherapy. To be effective a counsellor needs to develop good listening skills and an empathic understanding of the individual. The term non-directive counselling has been borrowed from Carl Roger's Person-Centred Counselling. It has been much debated (Clarke, 1991) and it is used as an ideal in genetic counselling to protect and endorse the individual's right to make an autonomous decision about personal testing.

The material presented in this book is based on the experience of working as a Psychiatrist/Psychotherapist in a Medical Genetics Department with clinical geneticists, genetic nurses and counsellors. It involved observing videos of clinical encounters, reading transcripts, personally conducting clinical interviews and providing supervision, consultation and discussion groups for the genetic counselling staff.

This book is written primarily for practitioners working in genetic counselling, regardless of their discipline and is a way of giving back to the world of genetics what actually belongs to it. However, the form is new, moulded into shape by using a psychotherapeutic understanding of communication competence and underpinned with the theoretical ideas of attachment theory. This provides a framework for thinking about behaviour, psychic pain, grief and the sense of personal narrative. To complete the picture the space between communication competence and attachment theory is filled with particular interview techniques. In so doing, clinical material and theoretical points will be plaited together to provide a balance between practice and theory.

The structure of the book can be thought of as paralleling the genetic interview, or the counsellor's professional development, in that in the early stages there are a lot of facts to be presented and understood. This can be very hard work. However, as the genetic counsellor knows, the facts are necessary to be able to progress and practise effectively.

The body of the book contains many case examples which are used to illustrate a theoretical point. To protect the confidentiality of the counselling sessions, the examples are fictitious in part, but based on actual clinical encounters.

The opening chapter provides an overview of genetic counselling, placing it in the context of developments in science, society and the individual. It explores the nature, range and components of a consultation to understand its function. This is considered to be more than an exchange of technical information as it is a reflection on an important aspect of life. The interview has evolved to help the patient address the psychological tasks required when they have genetic information. In a consultation the individual is helped to self-reflect and consider what is involved in asking a genetic question and, as a consequence, is being prepared for the effects of a risk assessment or a test result.

The thesis of this book begins to develop in Chapter 2 where genetic counselling is reframed as a psychological stress. This gives genetic counselling a theoretical basis which explains the processes involved and the difficulties individuals encounter. Stress and coping go hand in hand and both are influenced by the interaction between the individual and environmental factors. As a result, there is a range of differences in perception, approach and reaction to genetic counselling with the individual differences categorised as mature, defensive and symptomatic.

In Chapter 3 these individual differences in stress response and coping behaviour are explained by using the framework of attachment theory. This theory explains how feelings are managed differently by individuals depending on their early attachment experiences to a caregiver. In addition, the theory explains other differences and these relate to the construction of a personal narrative, the ability to self-reflect and use constructive thinking processes and includes the nature of the relationship established with the counsellor. The theories of stress, coping and attachment behaviour not only provide a framework for understanding individual behaviour, but also establish a framework which informs the counsellor's response.

Chapter 4 discusses the role of the counsellor who has the complex task of providing factual genetic information whilst also acting as a facilitator in the decision-making process. As the chapter progresses, the theme is built up of the counsellor helping the individual to prepare for decision-making by assisting in the processing of unresolved emotional issues from the past and present. This promotes a self-reflective approach to the counselling process. The principal skill of the counsellor is empathy which, by connecting the counsellor to the individual, results in the containment of anxiety such that chaotic experiences can be thought about.

Chapter 5 presents examples of how the counsellor works to help the individual contain anxiety, modify defences and attachment patterns to facilitate thinking and coping. It demonstrates the importance of resolving the psychological issues for the individual to be able to consider and absorb genetic information.

The idea of a system is introduced in Chapter 6 as the counsellor not only works with individuals, but also with a family group. The family is a social system made up of individuals of different generations, where the individuals are the inter-related parts of the system which mutually affect one another. Systemic thinking is not only confined to working with the family as the counsellor also thinks systemically in an individual consultation. The chapter links systemic thinking to the attachment theory presented in Chapter 3 and highlights that the motto of a secure family is 'collaborate to care'. The central issue for families is how they organise themselves around information about their genetic inheritance. The discussion includes looking at how information about a genetic disorder could be thought of as another element which needs to be included into the family system in the same way as a birth or death triggers an adjustment relating to the life cycle.

Chapter 7 focuses on examples of working with families and children. It alerts the counsellor to the presence of children in the consultation and how an understanding of family structure and child development can be an asset. It includes examples where there is a particular request for testing of children for adult-onset disorders.

Chapter 8 explores the influence the nature of the disorder has on the consultation by exploring four specialist areas: prenatal problems, Huntington's disorder, cancer and dysmorphology. The nature of the disorder and the implications for the individual, especially with regard to future treatment or

monitoring, affect the behaviour of the counsellor in respect of the balance between the educative and psychological interventions.

In Chapter 9 the effect on the counsellor is explored and discussed under two headings: the issues which are innate in addressing genetic problems and countertransference issues. The latter refers to all the feelings evoked in a counsellor as a result of working with a particular individual. It consists of issues relating to the counsellor's personal life and those triggered by the individual. A simplified version of countertransference is presented which is based on the individual's attachment pattern and management of processing emotions, as well as undeclared aspects of the individual's thinking or feelings which are projected into the counsellor.

The final chapter (Chapter 10) returns to the idea at the beginning of the book. It takes an overview of genetic counselling, not exploring the nature or component parts but rather focusing on the attitude within the consultation. It debates the concept of non-directiveness. Historically the concept was developed to support individual autonomy in decision-making, but it has been misinterpreted and sometimes it has been turned into non-involvement and passivity or even coldness. The chapter draws on the work of a number of authors in family therapy and psychoanalysis and traces the development in their thinking of the concept of neutrality. It includes the concepts of curiosity, narrative therapy and the counsellor's position of 'not-knowing' where there is no fixed answer, but a number of different perspectives.

1

An overview of genetic counselling

It is tempting to begin this book about the psychological aspects of the speciality of medical genetics by focusing on the individuals and families concerned and to explore the effect or the personal meaning of having a genetic consultation. However, good medical training and, in particular, training in psychological principles begin with a wider view. It uses a wide-angled lens, rather than a more detailed focus, to explore and define genetic counselling. This means beginning by addressing how genetic counselling has developed into its present shape and looking at the context of its evolution. This sets the scene which is the context of the question of the nature of genetic counselling and how is it defined. This can be followed by looking at the range of the speciality and the motivation for seeking genetic counselling. With that back-drop, it is then possible to analyse the component parts of a typical genetic counselling encounter, discuss the function of the interview and ask why it takes its present form.

The context

Genetic counselling has evolved in the context of three different areas: advances in medical knowledge, changes in society and the basic human desire to have knowledge, to understand and to learn.

Advances in medicine and the study of diseases have progressed by refinements in clinical diagnosis and special investigations. The richness of knowledge about the factors involved in the development of diseases has necessitated divisions into specialities, which all come under the umbrella of medicine. The broadest division is into the internal or individual constitutional factors and the external or environmental factors of a particular disease. Scientific enquiry into the individual factors has resulted in identifying the gene