Genetic Transparency – Transparency of Communication

Gabrielle M. Christenhusz, Lorraine Cowley, Tim Ohnhäuser, Vasilija Rolfes

Introduction

This chapter will focus on ethical issues concerning the communication of genetic transparency. Elsewhere in this book there is a chapter devoted to defining what might be meant by genetic transparency,¹ but in this section we use the term broadly to indicate what can be known about a person’s genes, by whom and for what purpose, in three very specific contexts: non-invasive prenatal testing (NIPT), pursuit of genetic diagnosis by parents of children with suspected/potential genetic conditions, and genetic testing for a known cancer predisposition.

In each context we will discuss not only what can be known within the technical limits of new and existing technologies (the extent to which our genetic transparency can be known) but also what meanings of genetic transparency can be or might be attributed to, derived from or assimilated into our cultural, social and working practices.

The first two sections of this chapter focus on communication between a medical professional (in our specific contexts, a gynaecologist and a clinical geneticist) and the parent/s (specifically, the pregnant woman or the parents of children with suspected/potential genetic conditions). The final section of the chapter focuses on communication between family members who have made a choice for or against predictive cancer testing. In all

¹ See ch. 1.
cases, we understand communication to be multi-directional, flowing back and forth between the various communicating partners. This communication may encompass both technical aspects (e.g. the genetic test’s analytical and clinical validity and clinical utility) and questions of meaning. It is important to acknowledge that various factors may hinder or challenge the communication process. From the patient’s point of view (and here we also include parents and family members), these factors will include literacy, personal and family history of illness and loss, recent traumas, and risk tolerance including the availability of social and clinical support. We also note that the various reactions of the family to personal and family experiences of disease and loss are more important than the mere fact of having these experiences; for example, childhood epilepsy may be a shameful trauma in some families, and just a fact of life in others. This leads to a sort of “phenotype-meaning gap”, analogous to the genotype-phenotype gap (one genotype can lead to diverse phenotypes, and diverse genotypes can lead to a single phenotype): here, particular phenotypes will be interpreted differently by different people. Thus we acknowledge that those who are communicating with each other may find themselves talking about different things while expecting to be understood. It is from this briefly stated understanding of some of the issues surrounding communication that we approach our case studies in this chapter.

The cases we use to illustrate our discussions are from the contexts of Germany, Belgium and the United Kingdom. The way in which genetic counselling is offered is different in each of these countries; in Germany and Belgium, for example, genetic counselling can be offered by non-specialists such as gynaecologists or other physicians and the specific profession of “genetic counsellor” does not exist, whereas in the UK it would be offered mostly via the genetics service comprising geneticists and genetic counsellors. Furthermore, the regulation of genetic testing across these countries varies. For example, the current German and Belgian models of service provision are self-regulated, while the UK model is

---

2 Sanderson/Zimmer et al., 2005.
3 Rolland 1987; Christensen and Green 2013.
4 Christenhusz/Devriendt et al., 2015.