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Project Synopsis

CESAGen: Genetics, Health and Identity

Professional and parental autonomy and involvement in childhood genetic testing

Established in October 2002, the ESRC Centre for Economic and Social Aspects of Genomics (CESAGen) is a Lancaster-Cardiff collaboration (Prof. Ruth Chadwick as Director, and Prof. Paul Atkinson as Associate Director). CESAGen's objective is to work with the relevant genomics science whilst attempting to clarify the human (social and economic) factors which shape these technologies and systems of knowledge. HCRC is an active participant in this programme of research.

In recent years the expansion of the possibilities of genetic testing have revealed clear differences in the ways in which both families and professionals approach the genetic testing of children. Testing is appropriate, and indeed necessary, in contexts where it may achieve a diagnosis for an already sick child or lead to a helpful medical intervention. Identifying carrier status for a recessive disorder or a chromosomal rearrangement is unlikely to be of any relevance to the child's own ongoing physical well-being but may have significance for reproductive decisions for the future adult. This aligns with the view that predictive testing of currently unaffected individuals for late-onset familial disorders is not essential but may be desirable.

The consequences of childhood genetic testing are as yet unclear. It is feared by some that testing may damage a child's future autonomy, may breach confidentiality and may have possible psychosocial ramifications for the child personally and in his/her ongoing familial relationships, but experience is limited.

This research will investigate how parents (and professionals) reach a decision to test or not to test a child. It will examine the explanatory resources that inform such a decision and identify how the selective use of clinical information, ethical and legal considerations and family-relational influences impact on how a decision is made and who is included in the decision-making process.

The study will examine data on a wide range of disorders for both predictive and carrier testing for children under sixteen. Our primary data will be gathered through semi-structured interviews with families who are considering genetic testing and also with 'retrospective' families who have previously decided whether or not to test. We will complement this material with data drawn from genetic counselling sessions.

We are interested in the following set of questions:

- How do parents account for their request to arrange genetic testing on their child, or their decision not to proceed with such a test? In what ways, if any, do the accounting practices differ in the two contexts of predictive testing for late-onset disorders and carrier testing?
- How do parents and professionals come to a decision about whether or how to involve a child in the decision about genetic testing? What influences (e.g. ethical, legal, clinical, family-relational) are foregrounded or backgrounded in these accounts?
- What evidence is there to suggest that the selective genetic affinity that may arise from childhood genetic testing disrupts close family units and their ongoing scripts?
- What evidence is there as to whether knowledge of their genetic constitution – their genetic identity – may challenge or reinforce an individual's other personal, family-relational or social identities?
- What is the impact of genetic knowledge on an individual's personal and familial identity? How do individuals accommodate to, assimilate or reject genetic information?
- What genetic information do parents transmit to their children and when/how is this done?
- How is the issue of responsibility for the transmission of genetic disorders managed in the family? When is this process implicit and when explicit?

This research, by documenting the perspectives of family members, will give voice to members of families with a range of genetic disorders, and will contribute to existing knowledge on family communication about genetic disorders. We hope that it may, in a modest way, strengthen their representation in policy debates. We also hope that our research findings will explicate the public and private processes of decision making around genetic testing and assist the development of clinical and counselling practices.

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