The Eugenics Stigma: The Role of the Genetic Counsellor in Prenatal Testing

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The National Society of Genetic Counsellors (NSGC) defines Genetic Counselling as the "process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease".

Genetic counselling and the association of a negative eugenics stigma is a complex issue. As genetic counselling combines genetic information with reproductive choices, some writers have drawn parallels to eugenics. This poster presents the findings of the literature review undertaken in order to understand the role of genetic counsellors in complex termination cases where eugenics can play a role.

Case Study

This case study forms the inspiration for this analysis of genetic counselling and prenatal testing.

- Young couple 15 weeks pregnant with their first child with confirmed trisomy 21, Down syndrome.
- Routine ultrasound showed soft markers for Down syndrome were found including a cystic hygroma and absent nasal bone.
- Genetic testing: First trimester CVS with CMA and sequencing, was subsequently undertaken and Trisomy 21 was confirmed. Several weeks after a CGH array was conducted and Trisomy 21 was re-confirmed as well as the discovery of duplication on 1p15.5.
- Parental CGH array found that the unaffected father also had 1p15.5 duplication (double the amount of DNA).
- Mother had previous pregnancy ending in miscarriage at 18 weeks (first trimester).
- The father wanted to terminate the foetus whereas the mother did not due to religious convictions.

The conflict evident between the parents raises issues of termination and whether it should occur following prenatal testing. As the time of writing, no decision had been made in regard to termination.

Phenotypic Dependence and why people terminate

- The role of how a disease presents is an area which genetic counsellors play a role in how the role of disability is framed in discussion. There are numerous aspects in which this phenotype changes people's minds.
- The complexities of eugenics is evident in the influence on families' chances of phenotype variant, with the key tipping point being Intellectual Disability, referred to as mental retardation.
- Wertz et al. (1991) presented data concluding that women were 94%–95% more likely to have an abortion when presented with severe mental retardation, including Down syndrome, were typically diagnosed.
- 94% of terminations were conducted following the discovery of lethal anomalies or conditions, which resulted in "substantial physical, mental disabilities, or both." (Pergament and Pergament, 2012).
- The unknown phenotype resulting from the 1p15.5 duplication complicates the case study further - quality of life.

Multifactorial Influence and why people terminate

- The reason chosen to have termination comes from a variety of sources and it is important that genetic counsellors respect this.
- The exploration into modern eugenics is found in journals where ethicists, doctors, nurses, the public, and geneticists have debated the topic for years.
- The beliefs of the patient and the family must be considered of higher importance.
- Religion, the media, individual and familial experiences and other factors including emotional and social pressures influence beliefs.
- Families have the final call in decisions; yet understanding their thoughts helps aid the genetic counselling and the discussions surrounding options.
- Acting as a professional, genetic counsellors' opinions are not presented to the patient due to "non-directive counseling".

The issues of eugenics in genetic counselling is one of multiple competing factors that cannot be prioritised but rather viewed as a whole that creates a unique picture for each individual, and

Pre-Implantation Genetic Diagnosis and eugenics

- PGD provides a new avenue of genetic counselling and options for couples to avoid certain conditions and therefore a new controversial area for the profession to be aware of.
- PGD involves completing genetic testing prior to embryo implantation through IVF methods; although this technology is still not perfect, with pregnancy rates at less than 50% as of 2012 and limited accuracy in terms of genetic analysis (Pergament and Pergament, 2012).
- In rare recessive conditions, a very large proportion of the mutation alleles are held in the heterozygous condition as "carriers".
- Harp-Weinberg equilibrium, alleles are found in a set genotypic frequency across generations regardless of whether presented in the dominant or recessive state (Hardy, 1908).
- Individuals who are carriers for autosomal recessive conditions are not implanted at the embryonic stage, removing their mutation allele from the population frequency as presented in the heterozygous state.
- By removing carrier status embryos translates directly to eugenics as mutations can be removed from the population.
- King's (1999) emphasis on "purpose and outcome" in declaring the processes behind eugenic reiterations how PGD is incorporated.
- End couples initiating PGD with the purpose of removing individuals affected with genetic conditions and carrying through that outcome, does this is eugenics.
- PGD removes the need for abortion and the moral issues that surround it, thereby providing a real alternative to many people.
- Australia's legislation in relation to PGD, set out by the National Health and Medical Research Council (NHMRC), states that PGD can only be used for detection of "serious genetic conditions [and] to improve an individual's chance of having a child with a disability" (NHMRC, 2007); yet there is no definition of "serious".

PGD is the strongest examples of eugenics in modern clinical genetics and the role of the genetic counsellor cannot be underestimated. Despite a clear scientific definition of eugenics, it is apparent that there is more personal element and the patient role is key.

Final Thoughts

I went into this project with the assumption that I would come out of it with a clear understanding of the issues at hand in modern genetics and the associated variables found within eugenics and prenatal testing, but more importantly, I would have a clear conclusion. This is not the case. The context behind the issues presented are far more complex than I ever hoped to outline.

Consequently, I wish to conclude that it is every individual's right to choose following the presentation of the facts involved. The role of the genetic counsellor themselves cannot be over emphasised and forms the foundation of the process I have proscribed here.

We all draw the line of eugenics at a different point, for some the gender segregation of prison counts as eugenics because we are attempting to stop criminals from reproducing. For others its not eugenics unless we are removing certain conditions from the population by force.

I have scratched the surface on the issues faced by genetic counsellors but hopefully, I have been able to help even a little bit in regards to the process by which Genetic Counsellor should approach the problem that has come to define the field. We should not be changing the minds of the patient, but rather educating genetic counsellors on the significance of the role they play and the information a decision reached purely by the couple based on their unique circumstances.