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A factor-finding search based on decision making surrounding Preimplantation Genetic Diagnosis in the United Kingdom

Research Summary and Findings

My research aimed to explore the extent to which attitudes surrounding genetic disease and disability influence decisions towards Preimplantation Genetic Diagnosis (PGD) in the United Kingdom (UK). Particular attention was drawn to diseases which vary in their clinical symptomatic manifestation, and are therefore considered to be 'controversial', as such diseases are being increasingly licensed by the Human Fertilisation and Embryology Authority.

The findings from my research detail several inter-related factors which materialised from the interviews for this study and are argued to shape perceptions of how severe a disease and/or disability is considered to be. These inter-related factors, influenced by humanitarian values, are reasoned to impact moral judgements on the social acceptability of risking the transmission of controversial genetic conditions to future generations. The insights gained from my research contribute to the understanding of factors which both drive and impede decisions to use PGD, and further suggest how perceptions of PGD may alter with the hypothetical introduction of genetic editing.

Brief Background

In Vitro Fertilisation (IVF) is a technique through which an egg is fertilised by sperm outside of the body (HFEA, 2014a). PGD is a two-stage process branching from the platform of in vitro fertilisation in which externally created embryos can be tested for specific genetic diseases before they are transferred to a woman's uterus (HGC, 2004). The process requires a biopsy of one or two cells to be removed from the eight-cell embryo. These cells are then analysed for whichever gene is known to cause the inheritable disease (HFEA, 2014b).

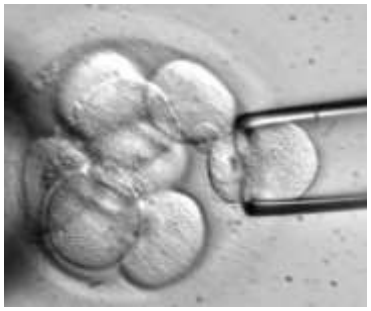

Image 1: PGD Biopsy (IVFMD, 2017)

Figure 1: PGD Analysis

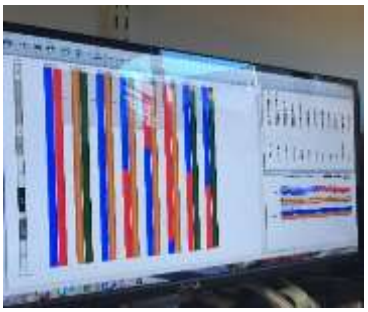

Figure 2: PGD Results

There are currently around 107 IVF clinics in the United Kingdom (UK), only twenty-two of which are licensed to carry out PGD (HFEA, 2017a), and each of these clinics differ in what genetic diseases or combination of diseases they are licensed to test for (HFEA, 2017b). Clinics are licensed by the HFEA which is an independent regulatory body that was created in 1991 following the introduction of the Human Fertilisation and Embryology Act (HFE Act) in 1990 (House of Commons, 2007: 16).

In addition to licensing fertility clinics and centres involved with treatments such as IVF and PGD, the HFEA is also responsible for overseeing the regulation of gametes and embryos used for fertility treatment and research (HFEA, 2017c). The HFEA was established as part of the HFE Act 1990 to ensure that the limitations set by the Act are adhered to without bias, to appease society at large, and as an avenue to safeguard new and ongoing developments in the field of human fertilisation and embryology (Mulkay, 1997).

Inherently, PGD is only made possible through advances in knowledge surrounding genetic diseases, and is reliant upon information from identifiable genes associated with a given disease to be utilised. Currently there are only around 420 diseases that are licensed to be tested for via PGD, with roughly a further 20 licences for testing pending (HFEA, 2017b). However, theoretically, PGD could essentially advance to prevent the transmission of 10,000 genetic diseases to future generations (should licences be granted) as research unfolds (Plows, 2011: 100).

With the ongoing expansion of genetic innovations, knowledge and services, individuals/couples do not necessarily have to arrive at PGD following traumatic or painful experiences (Kerr, 2004; Franklin and Roberts, 2006). The increase of services available mean individuals/couples with a genetic disease can choose not to risk transmitting their disease to a child without having necessarily lived with adverse clinical symptoms of the disease themselves (House of Commons, 2005). However, unlike the conclusiveness of ‘serious’ conditions, many of the controversial conditions now licensed are deemed so because there is recognition that even if the child inherits the disease they may still be able to live a high quality of life.

In the UK, the criteria for PGD is refined specifically to medical and therapeutic purposes. Thus, despite misconceptions, PGD in the UK is largely a choice which applies knowledge to prevent genetic disease, and an opportunity to improve the health of future generations by doing so (Kerr, 2004: 4; Franklin and Roberts, 2006). PGD in the UK cannot legitimately be used for social reasons or to select for non-medical characteristics (Birk, 2009).

Participants

Participants for my research were largely recruited through an online support group on Facebook and via the charity Genetic Disorders UK. The final participant sample consisted of 15 women and 2 men, aged between 21 – 50 years. The majority of the

participants have a musclo-skeletal condition called Klippel-Feil Syndrome which is often associated with other diseases and/or disabilities such as scoliosis, spina bifida, and torticollis (NORD, 2016). As such, several participants presented with more than one disease and/or disability. Other diseases within the sample included Multiple Epiphyseal Dysplasia, Lupus, and Neurofibrosis.

Interviews with my participants were conducted between 17th December 2016 – 2nd March 2017, and lasted between 24 - 45 minutes. Due to the modest size of the sample used for my research, findings are indicative and may not be representative, thus cannot be wholly generalised to the population at large but are transferable. To protect anonymity participants were assigned a pseudonym, and their pseudonyms have been used in this research summary, as well as any other publications relating to my research.

I remain grateful to all my participants who dedicated their time, life experiences, and opinions to participate in my research. My research would not have been possible without my participants' generosity and understanding.

Findings

Unsurprisingly, my participants envisaged PGD as an option to prevent the transmission of disease from parent to child and understood this intervention to be its legitimate primary purpose**.** However, in consistence with findings from other research on PGD (Franklin and Roberts, 2006; Plows, 2011) my research found that the main aim of utilising PGD was not necessarily to eliminate the incidence of disease from society, but an expression of a sense of moral obligation to prevent avoidable pain and suffering. As Matthew explains:

> *they don't want their child to suffer from the same genetic disability […]*
> *they don't want their child to go through the same pain and suffering*
>
> (Matthew, 39, Investment Banker)

Pain and/or suffering were discussed in both physical and psycho-social terms, as well as being perceived as something that would have a detrimental knock-on effect to most areas of life. 'Pain' and 'suffering' were repeatedly expressed to be at the heart of what it meant to have or live with a disease and/or disability. Participants in my research cited psycho-social struggles, various medical interventions, and/or dependency on others as factors they desired to avoid transmitting to their hypothetical children and future generations. The latter two were often pivotal to the management of participants' ongoing symptoms and pain, yet they were expressed with an air of misfortune. Collectively, the three factors formed the basis for most considerations towards decisions surrounding PGD throughout the interviews with my participants.

My participants' perceptions pertaining to quality of life were closely linked to the severity of a disease/disability, and it was these perceptions which greatly influenced attitudes towards utilising PGD. Cross-analysis of the interviews revealed that my participants determined the severity of a disease/disability by the combined pain and adversity which could result from the discussed factors, in addition to life expectancy.

> *[Serious would be] something which is life threatening or life changing. Gives a lot of dependency, or reduces life expectancy. Life changing would be living in a lot of pain or not being able to function normally or be able to have a proper independent life*
>
> (Beth, 42, Ex-Accountant)

Palpably, because what is considered to be adverse for some is not for others, discrepancies in what is 'severe' or 'serious' add to the controversy which surround some of the genetic conditions now licenced for PGD. Many risks, symptoms and characteristics associated with many diseases and disabilities sound abhorrent, and whilst individuals may not be able to comprehend possessing the ability to deal with such circumstances physically and/or emotionally given the situation, individuals can learn to adapt (Livneh, 2001; Marteau and Dormandy, 2001). My participants concurred that their life experiences of living with a disease and/or disability can be less detrimental to quality of life, and much more manageable than lay perceptions perceive them to be.

Over half my participants believe knowingly transmitting a disease and/or disability which was at risk of being highly debilitating[1] to be wrong and/or unethical. I believe some of my participants held this judgement because actions relating to knowingly transmitting a genetic condition deviate from aiming to generate a future with greater levels of wellness. The possibility of another generation experiencing the '*same*'[2] adversities as the parent is what my participants considered to be immoral.

Interestingly, perhaps because of the controversial nature of my participants' genetic diseases, and/or many of my participants believing they have a good quality of life despite pain and adversity experienced; many of my participants shared that their personal desire to create and have a child would have overruled consideration to the judgements outlined above. Whilst my participants expressed wanting to prevent pain and suffering in relation to the interrelated factors as a moral obligation, facilitation of this obligation was prominently overturned when it came to my participants' individual choices and decisions:

> *I'd put myself at risk to have a child, because I've always wanted a child so I would do anything to have a child*
>
> (Laura, 30, 'Never worked')

Participants in my research who were willing to risk transmitting their genetic condition to their child communicated that they were mindful of the uncertainty which

[1] My participants determined the debilitating distinction of a disease/disability by how the combined culmination of factors detailed in the opening findings present.
[2] See Matthew's quote on page 3

surrounds the manifestation and presentation of controversial diseases, including their own genetic disease(s), and this possibility was something they had often contemplated at length. Participants who already had children admitted that had they been diagnosed with their respective genetic conditions procreating, their reproductive decisions would not have changed to fulfil the moral responsibility they had previously conveyed. Though, it is recognised my participants were speaking hypothetically, and as such, perceived behaviours may not match actual decisions:

> *I didn't know I had it before I had my children, but knowing that I have it now still wouldn't have stopped me because I wanted children*
>
> (Rachel, 34, Part-time Student)

Nonetheless, my participants shared they would still undoubtedly love and care for any child that was born to them regardless of whether it had a disease/disability. This stance was true of participants who already had children, those who wanted to have children, and those with no plans for having any (further) children. In cases where my participants were willing to accept the risk of transmitting a disease and/or disability to create a child without utilising PGD, it is noted that they seemingly countered their internal dilemmas and concerns of doing so with hope. Hope that even if their child did inherit their condition prognosis determined by medical professionals would prove to be wrong.

Participants who had/knew children who have inherited a genetic condition, particularly a condition that has unfortunately manifested and presented to a greatly debilitating extent, were also rarely deterred from wanting to procreate. Such participants in my research however, bypassed discussing the child's pain, suffering, and abilities, and alluded to personality and character traits of the child. Participants seemingly felt the need to justify the value of these children, and defended the child's existence by the emotions they evoked in the those that love and care for them. Thus, a pricelessness, which would feasibly be readily advocated by many disability activists (Kerr, 2004; Kerr and Shakespeare, 2002), centred on humanitarian values as opposed to abilities, was repeatedly accredited to the children that participants in this study passionately commented upon.

To my understanding, my participants felt an unmistakable responsibility towards creating a better future. Thus, my participants were appreciative of PGD providing a welcome opportunity for individuals who without the intervention, may not otherwise choose to procreate. However, whilst some of my participants considered PGD a choice that some individuals may welcome, and commended the intervention for the benefits it could bring, PGD was also met with distaste from other participants in my research. Though a minority, just under half of my participants felt that the process of PGD was disrespectful of life. The fact that PGD still involves the necessity of selection, termination, and death, even if it occurs before the embryo is implanted to try and achieve a viable pregnancy (Franklin and Roberts, 2006: 157), made the intervention quite unpalatable for some:

> *I am not personally in favour of the IVF and wasting so many embryos, they are human and you want to have one healthy one on the cost of so*

> *many unhealthy or dead ones, let's say dead ones yeah because you would kill off so many, for your own pleasure really*
>
> (Olivia, 47, Physiotherapist)

Whilst other participants in my research did not consider embryonic cells to be as human:

> *test that early on an embryo, it is not seen as alive or a human being yet*
>
> (Annie, 37, Lawyer)

As such, the interesting distinctions in how my participants considered an embryo understandably influenced the extent to which they would personally consider PGD. Yet, a far more significant proportion of my participants felt that utilising PGD to prevent the transmission of pain and/or suffering was considered a moral obligation to society. Neglect to utilise PGD (if it is licenced for a disease) was a choice upon which my participants passed a distinguishable judgement. Although, it is noted that my participants' personal perceptions of their respective disease and/or disability in relation to their own life experiences undoubtedly influenced this consideration. Several participants in my research did not perceive their condition to be something that was adverse enough to require active, dedicated prevention, and as such their genetic condition had little bearing to their decisions surrounding procreation.

Additionally, whilst many of my participants expressed reluctance to utilise PGD themselves, they could easily comprehend why PGD may be sought and/or endured despite the expense, risks, and possible side-effects involved in the technique. In several instances, participants in my research understood PGD to be a wanted option because it does effectively allow individuals to attempt to create a child without the uncertainty of transmitting *their* known disease and/or disability. As such, perhaps in recognition to negative judgements that could be bestowed upon them, some of my participants cited an actual want for PGD, to relieve themselves from culpability, and to balance their responsibility towards generating a greater level of wellness with the risks of transmission.

Regardless of my participants' thoughts towards PGD in its current capacity, there was an unexpected consensus between participants in my research, towards PGD being the greater, more desirable, and morally superior option if genetic editing is added to its scope. Participants in my research who did not foresee using genetic editing technologies themselves, even held the potential of the hypothetical intervention in high regard. Reservations from some of my participants centring on right to life arguments with PGD in its current capacity seemed to diminish with the belief that genetic editing might enable a respectful intervention that could prove therapeutic to life, as opposed to destructive. Nevertheless, understandably, participants in my research still expressed reservations towards the use of genetic interventions, but these reservations were balanced with convictions on the good genetic editing could bring.

In continuation from the findings of this research, my forthcoming PhD research will explore the potential reproductive future with genetic editing to a greater extent.

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