Discuss the purposes of genetic testing during pregnancy and the ethical issues raised by such testing

Genetic tests during pregnancy are usually carried out in order to check for any abnormalities in the child itself, and to ensure the safety of both the foetus and mother. Genetic tests look mainly for the presence of mutations, chromosome abnormalities, or genetic disorders (Cunniff, 2004). Traditionally, the major focus was on chromosomal abnormalities (Lau and Leung, 2005), in particular Down's Syndrome. This procedure would involve looking at the chromosomes using karyotyping (Stanford et al, 2006). There have been major breakthroughs over the past few decades and more sophisticated tests now exist for a variety of different mutations, disorders and diseases during pregnancy. This has extended to tests before implantation as well, due to New Reproductive Technologies (NRTs). In order to carry out these genetic tests, cells are usually take from the foetus using invasive procedures. These procedures range from amniocentesis and chorionic villus sampling (CVS), as well as celocentesis (Lau and Leung, 2005), to foetal bloody sampling, biopsies and microarray. The procedures are potentially dangerous and could result in foetal loss. This danger is just one of the many ethical issues on the grounds of which people object to genetic tests during pregnancy. By looking at the spectrum of disorders and diseases that genetic tests can be carried out for during pregnancy and the implications of testing for them, to the implications of new technologies that are being created enabling tests for other traits as well, the essay aims to outline the purposes for such testing and the ethical issues raised by genetic tests during pregnancy. The topic will be expanded to look at one neonatal test, the Guthrie test for phenylketonuia to illustrate one of the main purposes of genetic screening, and pre-

implantation diagnosis, for example tests during in-vitro fertilization (IVF), as this is the real ethical battleground of the future.

Most pregnant women are offered a variety of screening tests and, where it is deemed appropriate, specific diagnostic tests as well (Human Genetics Commission, 2006). Prenatal diagnostic tests provide diagnosis of particular conditions the baby may have (Ibid), and are carried out when there is a 'familial, maternal or fetal condition that confers an increased risk' (Cunniff, 2004). Prenatal diagnostic tests will often be carried out if there are abnormal results to screening or other tests (Ibid). None of these tests, however, are entirely reliable (Human Genetics Commission, 2002), and whilst undergoing them it is important that the patients make decision in full knowledge of this fact.

Spina bifida is associated with certain chromosomal abnormalities, and can cause severe disabilities and death. Amniocentesis is used to obtain foetal cells from the amniotic fluid, which can then undergo 'fetal chromosome analysis' (Cunniff, 2004) to determine whether the child might have spina bifida. This is most commonly performed between fifteen and eighteen weeks of gestation (Ibid), and though there are risks of miscarriage, they are not high. Maternal serum screening and chorionic villus sampling may also be used for an earlier diagnosis at between ten and twelve weeks. Some clinical trials have indicated that chorionic villus sampling may result in a slightly higher foetal loss rate than amniocentesis, but 'earlier diagnosis provides additional time for counseling and decision-making' (Ibid).

In the United Kingdom, abortion is illegal after the twenty-fourth week of pregnancy, with certain exceptions. One of these exceptions is a 'substantial risk that if a child were born…it would be seriously handicapped' (Human Genetics Commission, 2006). Each year there are around 1, 900 terminations due to a foetus having a severe disability or handicap (Ibid). Spina bifida is considered one such serious handicap and a couple can choose to terminate a pregnancy on these grounds if they discover that their child will have it. Many religious groups disagree with the concept of aborting on these grounds as they consider it the will of God. It is often debated, however, that it is likely that spina bifida will result in death anyway, and it would be far worse if a child and parents had to live with the disability for several years before then suffering loss anyway. On the whole there is a general consensus that it is acceptable to allow these tests in order to give the option of termination, or even the option of treatment such as foetal surgery (Cunniff, 2004) though such treatments hold risks.

Cystic fibrosis is a hereditary disease due to over one thousand gene mutations. Up until now molecular tests were carried out using samples obtained through invasive methods and the polymerase chain reaction (PCR) to replicate the genes. These methods unfortunately are neither time nor cost efficient (Lau and Leung, 2005). Whole human genome microarray, a form of gene-chip technology, is another possible test for genetic defects. This allows comparisons between normal genes and those with a particular disease and may enhance prenatal diagnosis of diseases such as cystic fibrosis (Ibid). Advancements in gene-chip technology in the foreseeable future will allow the study of thousands of mutations in one experiment (Ibid), and thus the identification of huge

numbers in society who have or are heterozygous carriers of a specific disease or disorder. Though there are advantages in that genetic testing would take less time, and people would be able to take preventative measures and treatments, if the information that were gained through any one of the prenatal genetic tests outlined were available to insurers or employers it could be misused (Human Genetics Commission, 2002).

One of the purposes of genetic testing is to enable preventative treatment for certain conditions or diseases if an individual is aware that they have the genetic trait that predisposes them to it (Lau and Leung, 2005). Phenylkentonuria is an autosomal recessive disease caused by the absence activity of the enzyme phenylalanine hydroxylase (McConkey, 1993), which converts phenylalanine into tyrosine (Ibid). The disease can cause neurological damage and mental disabilities if not detected and treated. Fortunately, if the disease is detected an infant can be put on a special diet that prevents any serious or permanent damage, and allows them to live a normal life. Though it is possible to predict whether a child is likely to have phenylketonuria if it is present in the family, tests for it cannot be carried out during pregnancy. The concentrations of phenylalanine are normal even in foetuses that are homozygous for the disease (Ibid), because proteins pass across the placenta from the mother who can convert phenylalanine to tyrosine in her own liver (Ibid). Blood tests, known as the Guthrie test, are carried out shortly after birth, which detect the disease and allow for preventative measures and treatment. There is very little that could be considered controversial about the Guthrie test as it does not create any great risk for the child or mother, does not provide potential reasoning for abortion, and can be beneficial in preventing the disease from developing.

The only potential ground for ethical battle is over the possibility that if the information about an individual's test results were to be leaked and misused it could hinder their opportunities, and this could be argued in regard to any confidential or personal records.

Down's Syndrome, or Trisomy 21 as it is also known, is the result of a non-disjunction error resulting in a third chromosome 21. For women of thirty-five years or older, there is a greater risk of an abnormal number of chromosomes in the foetus (Cunniff, 2006), and samples can be taken through amniocentesis, CVS or blood sampling to test among older women or if a possible abnormality is detected through an ultrasound screening. Foetal blood sampling is useful in these circumstances, as it may be used for 'rapid fetal karyotyping' (Ibid). The risk of loss, however, is between 1% and 2%, making it more dangerous than amniocentesis or CVS and giving another reason for objecting to the procedure.

A child with Down's Syndrome will have mental disabilities, certain physical irregularities and will need a lot of care, but it is not a fatal disease and as a result there are debates about whether this is strong enough grounds for choosing to abort a pregnancy. People with Down's Syndrome can have jobs, carry out day-to-day activities and live happy, relatively fulfilled lives. In some cultures Down's Syndrome are not considered significantly different from any other person (Detwyller, 1993), and many argue that one of the main dangers in being able to choose whether someone with Down's Syndrome should live is that society may move towards a eugenic ideal of what individuals should be like and begin aborting on other grounds as well. There are considerations, however, about who will care for an individual with Down's Syndrome,

especially once the parents have grown older and are not capable anymore. Questions have been raised about whether it is fair to expect other relatives to step in and lend a hand. Despite disagreements over whether or not abortion should be permitted on the grounds of a foetus having Down's Syndrome, the genetic tests provide other functions such as preparing parents for having a Down's Syndrome child, and giving them support and education about what lies ahead. A small minority may disagree with having the tests at all due to the risk involved, but new, less invasive techniques are being created that may counteract this argument (Hahn et al, 2008). Tests such as that for Down's Syndrome have benefits in enabling a couple to make an informed decision and prepare for the child itself.

In September 2002 newspapers[1] reported that scientists had identified a gene responsible for babies born with a cleft lip and palate, Van der Woude syndrome. Following this there were debates over whether or not the condition was serious enough to allow prenatal tests to see whether a foetus had the gene, and for the parents to therefore have the option of termination on the grounds of serious disability. There has been a lot of controversy over this issue as Van der Woude syndrome does not impair an individual's mental abilities, and can be treated meaning there is no reason that an individual is not fully capable despite having it. In March 2005[2] the British courts decided not to prosecute two doctors who authorized an abortion due to a cleft lip and palate on the grounds that the child would be handicapped. This event resulted in uproar as debates surged over whether a fixable facial deformity is grounds for terminating a pregnancy, and if so what

[1] The Guardian, September 2nd 2002, 'Genes causing cleft lips identified', James Meek
[2] The Guardian, March 17th 2005, 'Cleft lip abortion done in good faith', James Meikle

does this tell us about the society in which we live in regard to physical appearance and aesthetics. Not only is there the worry about the removal of certain traits or features that are considered less desirable from the population, but there is also the worry that some couples may want abuse technology and genetic tests to ensure that their child has a specific disability. This has been brought into light recently in the case of deaf couples wanting to screen for and ensure that their child is also deaf (Lau and Leung, 2005), bringing into question whether people should be allowed to choose for such traits.

Technologies exist and are being created opening the possibilities of prenatal determination of the sex of a foetus, and therefore gender selection through abortion possible (van Balen and Inhorn, 2003). Though abortion is illegal, it is another trait that could be open to abuse by couples that want a child of a specific gender, which is particularly common in countries such as China with its one child policy. Technologies are also being created that will allow DNA weighted semen selection (Ibid), as well as the existence of IVF during which it is possible that preimplantation diagnosis and selection could take place.

Preimplantation genetic diagnosis during IVF can be done using blastomere biopsy of a couple of cells from an eight to ten cell embryo (Lau and Leung, 2005). Normal embryos can then be replaced in the uterine cavity to become a foetus. The cells taken and tested also give information about other inherited traits, such as physical features or intelligence, and there are worries that couples will at some point in the near future begin wanting to choose the embryo which contains the most desirable genetic information.

The possibility of being able to pick and choose features and traits creates great anxiety, as it opens up the potential for designer babies and emphasizes the common misperception that everyone in society should fit into a conventional idea about perfection. Another worry is that these tests will only be available to the wealthy, thus dividing society to an even greater extent in regard to wealth and creating a genetic elite within society. The ethics behind the pursuit of perfection is a worry for many, but most people are still so horrified by the idea of a Frankenstein-like scenario that it is unlikely to become common practice any time soon.

That this technology exists has brought to the foreground questions over the pressure to test for and possibly abort foetuses with abnormalities (Ibid), and the potential emotional problems that the tests may cause for pregnant women. Uncertainties have arisen over whether, when these technologies are commercially available, they should be on offer to all pregnant women or just those that ask for the tests. After all, the tests are intended to benefit the women and allow them to make informed choices, not make them feel pressured into feeling they must make decisions about whether they want a child who may have imperfections and deal with the consequences of what they decide. It is important, therefore, whilst embarking on advances in the field of genetic technology to have an infrastructure in place that can provide support to pregnant women, the child and families.

Genetic tests before, during and just after pregnancy are done for a variety of reasons, from abnormal screening results to the knowledge that a trait is present within a family.

These tests have several purposes. Initially, by testing for abnormalities they allow a couple that are going to have a child with a certain disorder or disease the option to choose to terminate the pregnancy. There are ethical issues over abortion generally and where it is acceptable to draw the line. Laws exist that aim to put controls over abortion, whilst health services attempt to provide support, counseling and information to the couple. Secondly, of the trait is treatable, genetic tests allow for preventative treatment from an early stage, or even foetal surgery if possible. Foetal surgery does hold risk for both mother and child, but it may be that by doing so this prevents any worse damage or danger in the future. Early testing during pregnancy can allow safer termination and treatment, but can also provide couples that decide to keep the child despite any defects the time to learn about what measures they must take and get used to the idea of it. Finally, if a couple is aware that an autosomal recessive disease is present in both their families, and they are using NRTs for assistance in having a child anyway, there is the possibility that they could ensure that the embryo they use does not have the allele for that disease. This possibility of selecting traits has opened up debates on the ethical issues of being able to select other traits as well and the effect this will have on society as a whole. Other ethical issues range from access to the information provided by genetic tests, to tolerance within society for those who have some form of disability, and the effects on the emotional stability of pregnant women who feel pressured to have these tests carried out. Due to the costs and amount of controversy surrounding new innovations, it is unlikely that certain technologies will be commercially available for some time. As for the tests that are currently used, it is indisputable that there are huge benefits to the pregnant woman, her partner and family, and the child itself in their

availability. Provided that laws are in place, and that the woman is able to make her own decisions, it is unlikely that the worries behind the more extreme ethical battles will become reality just yet. For the time being, it seems the purposes for carrying out genetic tests during pregnancy, as well as before and directly after, including the ethical reasons for having them, outweigh the ethical objections.

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