

BioethicsBytes Guides to Streamed Media

1. Preimplantation Genetic Diagnosis

Introduction to this resource

Welcome to the first *Guide to Streamed Media* being produced by the *BioethicsBytes* team at the University of Leicester. These guides are intended to offer busy lecturers and school teachers advice on the best video and audio clips for teaching about the science and ethical implications of new developments in biology and biomedicine. The advent of online news archives, and broader support materials for other programmes, makes it relatively straightforward to incorporate streamed media of this kind into Biology, RE or General Studies lessons. This teachers' resource is designed to offer a guided tour through some of the most readily applicable clips available on the internet. In addition to providing background material, we have also produced some 'off the shelf' worksheets for use in conjunction with the selection of clips.

Within the text there are embedded hyperlinks that are underlined and marked by a pointing-hand logo (☞). If you wish to follow a link, hover over the relevant words and press Control & Left Click with the mouse. Most of these recommended clips are currently being streamed on the BBC website (www.bbc.co.uk). The length of each clip is shown in brackets. In addition to the streamed media themselves, there are also links to reviews and articles offering background to the field.

There is probably too much information here, and too much repetition between clips, to justify using all of the recommended material with a group of students. It is, however, suggested that you watch (or listen) to all of the streamed media before deciding which to show. You may then elect, for example, to include one of the introductory clips to demonstrate the potential of Preimplantation Genetic Diagnosis in the detection of disease, and then one from the later sections to give a more detailed application.

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Introduction to Preimplantation Genetic Diagnosis

Significant, but controversial, breakthroughs in the field of Preimplantation Genetic Diagnosis (PGD) are being made on a regular basis. As such, PGD is frequently the subject of news reports and documentaries. Reading our [☞ Bioethics Briefing on PGD](#) is recommended, particularly for discussion of early applications of this technology which pre-date the availability of streamed news, and for fuller coverage of both the science and ethical issues surrounding the technique. A 2002 review by fertility doctor Peter Braude also offers helpful background details and can be accessed via this link [☞ Braude *et al* Nature Reviews Genetics 3:941-953 \(2002\)](#).

PGD was first used in 1989 to determine the gender of an embryo (Handyside *et al*, Nature **344**:768-770, 1990). A region of the Y chromosome was amplified using the Polymerase Chain Reaction (PCR) to select *against* male embryos, which may have been susceptible to an X-linked disorder. Since then, the technology has been used to screen for an increasing number of disorders such as cystic fibrosis, sickle cell disease, haemophilia, and spinal muscular atrophy. In principle any inherited condition can be tested, provided that the genetics of the abnormality is understood. It has also been used to look at chromosomal disorders.

More recently, PGD has also been used to select *for* a tissue-matched, or “saviour”, sibling. The aim is to use stem cells from the cord blood of the new child for transplantation on behalf of the older. It is also now permitted to employ PGD in the diagnosis of some diseases that are late onset (i.e. that develop in people during their thirties or older) even if they do not have full penetrance (i.e. not everyone with the genetic defect will go on to have the symptoms of the disorder). Additionally, there have been ‘social’ uses of the technology, e.g. for “family balancing”, that is to select the gender of the future child for non-clinical reasons (although this is not currently allowed in the UK, and therefore couples seeking this option pursue treatment overseas).

PGD is controversial. Long-standing arguments over the sanctity of life and the moral status of the embryo are compounded in discussion of PGD and broader *in vitro* fertilisation (IVF) technologies. On the other hand, is it better to allow parents to choose not to bring a child into the world if they know that it will have a certain inherited condition, particularly if they have already cared for an older child with the same condition?

Linked below are two videos from the BBC website. The first, from August 2005, was at the time of a public consultation on changes to the fertility regulations, and includes a general introduction to IVF technology. Interviews with Baroness Warnock (in favour of liberalising the laws) and Fiona Pinto (from Comment on Reproductive Ethics, against liberalisation) highlight some of the issues involved. Jayson Whitaker discusses his family’s situation; see Section 1.3, below.

The second clip is support material from the 2006 series *A Child Against All Odds*. It features fertility doctor Simon Fishel giving a very clear explanation of the current scope of PGD. Note: there is a discussion of the other programmes in the series on [☞ BioethicsBytes](#).

Video links

[☞ Fertility laws set for overhaul - BBC News, August 16th 2005 \(2min 38s\)](#)

[☞ A Child Against All Odds - BBC, 2006 Interview with Simon Fishel \(4min 27s\)](#)

A set of questions for use with these clips can be found on the next page (Worksheet 1.0)

Introduction to Preimplantation Genetic Diagnosis (PGD)

Fertility laws set for overhaul – BBC News, 16 Aug 2005



http://news.bbc.co.uk/1/health/4185000/newsid_4185000/4185004.stm?tw=bb&mp=r

1. How many babies are produced in the UK each year using *in vitro* fertilisation (IVF)?
2. Who was the first test tube baby and when was she born?
3. What changes to the law does Baroness Warnock want to make?
4. Why do pro-life campaigners object to this technology?
5. Why do some people dislike the term 'designer baby'?

Interview with Dr Fishel, *A Child Against All Odds* – BBC 2006

1. How is PGD done?
2. How long after fertilisation is PGD carried out?
3. Name some of the disorders that can be screened using PGD and how many others could be tested?
4. Apart from the diagnosis of disease, what other ways can PGD be used?
5. Why do some people get upset about the techniques used?



http://www.bbc.co.uk/health/tv_and_radio/bb/acaao_experts_fishel_16x9_bb.ram

1.1 PGD for the diagnosis of genetic disease

Retinoblastoma (RB) is a cancer of the retinal tissue at the back of the eye and can occur in both hereditary and sporadic forms. Inherited RB can affect one or both eyes, whereas sporadic cases only affect one eye. RB usually occurs in children before they are five years old. The cancer is caused by a mutation in the RB-1 gene, and is transmitted in an autosomal dominant manner (i.e. there is a 50:50 chance of a child inheriting the disease gene from a parent).

Retinoblastoma is not incurable; a range of treatment options is available, including cryotherapy, laser therapy, chemotherapy, radiotherapy or surgery, dependent upon the stage and position of the tumour. However, children who have been successfully treated usually have some damage to their eyesight, and although the success rate in treatment is high (95%), there is an increased risk of secondary malignancy in those children with the hereditary cancer. Further information may be obtained from the [☞ Retinoblastoma website](#).

Both videos chosen to illustrate this use of PGD involve the Donovan family. Angela Donovan had RB as a child and is blind in one eye. Her son Kieran inherited the condition but was successfully treated. Angela and her husband Louis sought, and received, permission from the Human Fertilisation and Embryology Authority (HFEA) to use PGD to ensure their next child does not have the disease. The first clip is from the BBC news in 2005 when the Donovans were seeking permission for the procedure. There is a brief discussion of arguments for and against PGD. The second video is rather longer, and includes a more in depth interview with the family. It can be reached both via the Health section of the BBC website, and as support material on the [☞ A Child Against All Odds](#) website.

Questions raised by the piece include:

- Should doctors have a free hand to decide what conditions can be screened?
- If we can eradicate disease, should we?
- Should we just look at diseases that are lethal in early childhood? What about those, such as retinoblastoma, that can be successfully treated, or those that have a late onset?

Video links

☞ [Woman 'can screen embryos for cancer' – BBC News, August 18th 2005 \(2min 6s\)](#)

☞ [The 'right' to choose a healthy child – BBC Health \(7min 52s\)](#)

PGD for the diagnosis of genetic disease

- **Woman 'can screen embryos for cancer' – BBC News, 18 Aug 2005**
- **The 'right' to choose a healthy child – BBC Health**



http://news.bbc.co.uk/player/nol/newsid_4160000/newsid_4164600/4164600.stm?bw=bb&mp=1m#http://news.bbc.co.uk/1/hi/help/3681938.stm
http://www.bbc.co.uk/health/tv_and_radio/bb/scao_stories_donovans_16x9_bb.ram

1. What disorder is carried by members of the Donovan family?
2. How has it affected them?
3. Why does the mother want to screen her embryos?
4. What other conditions might this technique be used to detect?
5. Why is there debate over whether PGD should be used in this case?
6. Are the family right to choose to screen embryos even though there is effective treatment for RB? Why/why not?

Thinking deeper

- Think of three reasons why people might object to using PGD to screen for retinoblastoma?
- What are some of the practical issues for someone considering PGD (e.g. concerning health and finance)? Are there any wider implications for society arising from these issues?
- Who should decide which diseases can be screened?

1.2 PGD for the diagnosis of late onset disease

Many types of cancer do not develop until later in life and may require a combination of genetic and environmental factors to trigger illness. These raise different questions concerning the use of PGD since carrying the gene alone does not guarantee that cancer will result (in such cases the disease is said to have ‘incomplete penetrance’). The videos below give two examples of where PGD is used to diagnose cancers that are found in an older population, these are breast cancer and colon cancer.

There are about 40,000 cases of **breast cancer** diagnosed each year in the UK and of these about 5 to 10% are thought to be caused by a mutation in the ‘breast cancer genes’ BRCA1 and BRCA2. Breast cancer is classed as a common type of cancer with 1 in 9 women developing it during their lifetime. The risk of getting sporadic (i.e. not inherited) breast cancer increases with age, with more than half occurring in woman over the age of 65 ([☞ Cancer Research UK website](#)).

The proteins encoded by BRCA1 and BRCA2 are involved in the repair of DNA that may have become damaged as a result of natural or environmental causes. Researchers have found many mutations in BRCA1 and BRCA2 that give rise to an increased susceptibility to breast and ovarian cancer. If mutations are found in BRCA1 and or BRCA2 the risk of developing breast cancer is 60% to 80%. These inherited forms of the disease also tend to occur at a younger age than the sporadic forms. In high risk individuals with a strong family history of cancer, the removal of the ovaries, post child-bearing, can reduce the incidence of breast cancer by 60% and ovarian cancer by 96%. Some individuals may also choose to have an elective mastectomy to reduce the risk of breast cancer.

Link to breast cancer video

☞ [Cancer gene checks for embryos debated – BBC News, August 12th 2005 \(1min 47s\)](#)

The next video describes the case for and against screening for **bowel cancer**. Colorectal (bowel) cancer is the third most common type of cancer in men and the second most common in women. As with breast cancer, most cases occur in people who are over 40 years old, with the majority of patients being over 60 years old. Each year about 30,000 people are diagnosed with colorectal cancer but, if caught early, it can be treated successfully ([☞ Cancer Research UK website](#)).

No single gene disorder is the cause of all types of colon cancer, although in certain cases there may be a family history of the disease due to an inherited mutation. For example, familial adenomatous polyposis (FAP) accounts for 1% of cases of colorectal cancer. FAP is caused by a mutation in the APC gene, which codes for a so-called ‘tumour suppressor’. If the protein function is lost due to mutation in the APC gene, further mutations arise making it more likely for cancer to occur. If an APC mutation is present then the likelihood is that cancer will develop at a younger age ([☞ Cancer Research UK website](#)).

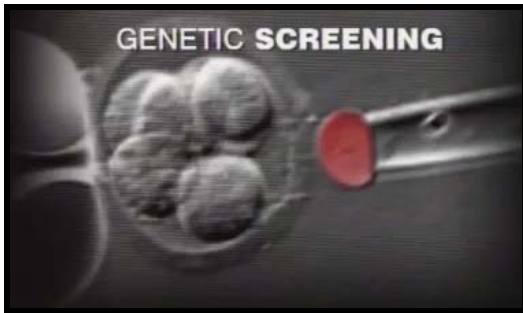
In the case of FAP there is a 100% certainty that cancer will develop if it is left untreated. The condition usually develops during early adolescence when hundreds of polyps form in the colon. These start out benign, but become cancerous due to mutations in the DNA arising over time. 95% of people with FAP will have developed polyps by the age of 35 ([☞ Wellcome trust genome website](#)).

Link to colon cancer video

☞ [Embryos to be screened for cancer – BBC News, November 1st 2004 \(1min 56s\)](#)

PGD for the diagnosis of late-onset disease

Cancer gene checks for embryos debated – BBC News, August 12th 2005



http://news.bbc.co.uk/player/nol/newsid_4140000/newsid_4145700/4145786.stm?bw=b&mp=rm

1. What disease has affected several members of Julie's family?
2. What treatment did Julie have done to avoid having the disease herself?
3. At what age does this disease typically affect people?
4. Who regulates the laws over genetic screening and fertility issues?

Embryos screened for cancer – BBC News, November 1st 2004

1. What genetic mutation does Emma Stevenson carry?
2. Has she passed it on to her daughter?
3. Would she want her embryos screened?
4. Why do 'pro-life' campaigners object to this sort of intervention why?



http://news.bbc.co.uk/player/nol/newsid_3970000/newsid_3970500/3970571.stm?bw=b&mp=rm

Thinking deeper

- If a person won't suffer from a particular disease until they are thirty, or even older, does this change whether or not it is appropriate to screen embryos for that condition? Why/why not?
- Is the option to screen for a growing number of conditions a natural development or is it evidence for the slippery slope to eugenics (i.e. influencing the genetic make-up of the human race by selective breeding)? Decide what you think and then list three arguments in support of the *opposite* view.
- Many of the cancer genes that could be screened have 'incomplete penetrance' (i.e. not everyone with the gene will get ill). You may therefore be discarding embryos that won't actually have the disorder. Does that change the arguments? Why/why not?

1.3 PGD for the creation of a tissue matched sibling

Charlie Whitaker suffered from Diamond Blackfan Anaemia (DBA), a disorder giving him low red blood cell counts. In about 50% of cases of DBA there may also be other congenital defects (present at birth rather than acquired and may be a result of either genetic defects or other effects).

DBA can be treated with corticosteroids and blood transfusions. By the age of 6, Charlie had received over 100 transfusions. Bone marrow transplants are considered an appropriate treatment and can cure the haematological aspects of the condition. The best possible outcome when performing a transplant is to have a tissue-matched donor (☞ [Vlachos *et al*, Nature 27:381-386, 2001](#)).

Unlike some other blood disorders (such as some thalassaemias, where the genetic basis is well understood), DBA is a disease characterised by genetic heterogeneity (i.e. a number of different genes may be involved in the disorder). It is thought that there are at least 3 loci in the human genome where errors in the DNA that can lead to DBA. Although there is some evidence of a familial pattern of inheritance in a small number of cases, the majority of cases appear to be sporadic. At the moment; it is impossible to screen for embryos that have the disease (☞ [D'Andrea *et al*, Hematology 58-72, 2002](#)).

The Whitaker family wanted to screen their embryos to select a tissue match for Charlie. Because you cannot test for DBA there is no benefit for the new child and therefore, at the time, the HFEA decided not to allow this procedure. As a result, the Whitakers choose to travel to America to carry out the screening procedure. The act of travelling to another country where the IVF and PGD techniques are not bound by such stringent laws has led to the term 'Fertility Tourism' being used by the media.

For the Whitaker family, this course of action led to the arrival of Jamie who was a tissue type match for his brother Charlie. When Jamie was born, stem cells were harvested from his umbilical cord and were successfully used in a bone marrow transplant for Charlie.

Partly as a consequence of the Whitaker's experience and the increased success rate of screening embryos (in terms of embryo survival) the UK law has since been changed to allow PGD for tissue-matched siblings to be considered on a case-by-case basis. There are a number of criteria that need to be taken into account before the HFEA allow PGD in such "saviour sibling" cases. These include whether the condition of the affected child is life threatening, and whether all other possible sources of material have been explored. The intention is that only the cord blood should be used rather than bone marrow or any other tissue due to the implications of increasing the suffering for the baby produced by PGD to help a sibling rather than being of a benefit for itself. See the ☞ [press release](#) from the HFEA for further details.

[Link to video about the Whitaker case](#)

☞ [Brother's tissue 'cures' sick boy – BBC News, October 20th 2004 \(2min 15s\)](#)

PGD for the creation of a tissue-matched sibling

Brother's tissue 'cures' sick boy – BBC News, October 20th 2004



http://news.bbc.co.uk/player/nol/newsid_3760000/newsid_3761300/3761374.stm?bw=bb&mp=rm

1. What was wrong with Charlie Whitaker?
2. How has he been 'cured'?
3. Why did the authorities refuse to allow this treatment in the UK?
4. What do the media call the selection of a baby as a donor?

Thinking deeper

- Is it right to create a baby to save another? Why/why not?
- The Whitakers chose 'fertility tourism' and went to a country with different rules to get treatment for Charlie. Was this a right or a wrong thing to do? Think of arguments for and against this course of action.
- What might be some of the psychological effects on a donor sibling? Do you think these might change if the treatment fails? In what way?
- If it turns out that the older child subsequently needs extra help from the younger, e.g. an organ donation, should this be done?
- Decide whether it is better to decide who should get treatment by having a general rule or on a case-by-base basis. Construct an argument to support your position on this.

1.4 PGD for social selection

This last section discusses the use of PGD for a variety of other ‘social’ reasons, such as selecting the gender of your child (for non-medical reasons) or the potential to genetically engineer a child of your choice.

Sex selection is carried out in many clinics in America and elsewhere around the world, but is not currently allowed under HFEA rules in the UK. There have been a series of cases whereby people have questioned these rules. One prominent example involves Alan and Louise Masterton who wanted to have PGD to enable them to have a daughter. They have four sons and previously a daughter Nicole, but she died as a result of an accident. The Mastertons expressed a NEED to have another daughter but, due to a previous sterilisation operation, Louise would have required IVF to get pregnant. They argued that it was only a small additional step to undergo PGD and ensure that a female embryo was implanted. Furthermore, they argued that since they were self-funding rather than seeking the operation on the NHS, this should have an influence on the decision. As it turned out, the Mastertons were not granted permission. Their story is told in the first clip linked below, which raises a number of arguments for and against a procedure of this type.

The second clip picks up on a related issue. Gender selection is illegal in India, but ultrasound clinics up and down the country are willing to determine gender and perform abortions if the foetus is not of the desired sex (there is a strong cultural preference for male children). In one area of India, Morena, the ratio of male to female births is 10 to 7; it is the worst in the country. One woman interviewed had aborted three female foetuses even though she risked imprisonment. It is estimated that 500,000 females are lost each year, and the motives are not necessarily the financial burden of a dowry. Middle class Indians are increasingly making a ‘consumer choice’ to have sons not daughters. The government is trying to educate people and has put in place an incentive whereby it will pay for the education of one girl out of each family from primary school up to university. Note: PGD is *not* being used in the specific example discussed in this clip, but it could be. Would increased availability of PGD make the situation better or worse in a situation where illegal sex selection is already so prevalent?

Links to video about sex selection

☞ [Father argues case for child sex choice – BBC News, March 24th 2005 \(3min 43s\)](#)

☞ [India ‘loses 10m female births’ – BBS News, January 9th 2006 \(2min 34s\)](#)

In a distinct case, discussed in an audio clip (from Woman’s Hour), a lesbian couple that are both deaf have sought a deaf sperm donor to achieve a non-hearing child. Again, this specific example did not involve PGD, but other deaf couples in the USA have argued that it ought to be allowed for this purpose (e.g. Dennis C. (2004) *Deaf by design*, Nature **431**:894-896). The debate in this programme involves Baroness Emma Nicholson, MEP, who is partially deaf, and Prof Tom Shakespeare, Director of Outreach for the Policy, Ethics and Life Sciences Research Institute (PEALS), who is achondroplastic. A number of interesting ethical arguments for and against this procedure are presented.

Links to audio clip on deliberately having a deaf child

☞ [AUDIO: Designer Baby – BBC Radio 4 Woman’s Hour, April 10th 2002 \(11min 47s\)](#)

PGD for social selection

Father argues case for child sex choice – BBC News, March 24th 2005



http://news.bbc.co.uk/player/nol/newsid_4370000/newsid_4378700/4378717.stm?bw=b&mp=rm

1. What treatment were the Masterton family wanting and why?
2. Summarise their arguments in support of their case?
3. Why do the HFEA legislate against the use of PGD for this process?

India 'loses 10m female births' – BBC News, January 9th 2006

1. Why are fewer female than male babies being born in India?
2. Is this happening in poor communities, or more widely?
3. What are the implications for countries such as India and China where there are social pressures to produce male offspring?



http://news.bbc.co.uk/player/nol/newsid_4590000/newsid_4596100/4596174.stm?bw=bb&mp=rm

Thinking deeper

- In what ways is the Masterton's request for PGD for "family balancing" similar to what is happening in India, and in what ways are they different?
- What arguments can be made in favour of PGD for gender selection, and what arguments can be made for not allowing it?

Designer Baby – BBC Radio 4 Woman's Hour, April 10th 2002

- Is it right for a family to choose a child to fit into their culture, even if this involves the deliberate selection for a particular handicap? Why/why not?
- How might the child feel when it grows older?
- Is this more to do with what the parents want rather than what is best for the child?
- Does it matter if the child is 'engineered' as long as it is loved and cared for?
- Who has the right to decide what disabilities are acceptable and what are not?
- If this was permitted, what other 'social reasons' might be given for PGD?