

Fact sheet – Huntingdon disease



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@ Bristol

Population frequency

1 in 18 000

Pattern of inheritance

Dominant, caused by a single gene on chromosome 4. An affected parent has a 50% chance of having an affected child.

Symptoms

Symptoms usually develop when people are between 30-50 years old, but this is very variable. Early signs include involuntary movements, concentration and memory problems and mood changes. These get more severe later in the illness. Death occurs on average about 17 years from the onset of symptoms, but this is again highly variable.

Treatment

At present there is no cure for Huntingdon disease, but medication, speech therapy and special diets can help treat the symptoms.

Possibility of genetic testing

Genetic tests are available - they will tell you if you have the gene, but will not give an indication of the age at which you will develop symptoms. Prenatal testing is also possible.

Fact sheet – Sanfilippo syndrome



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Population frequency

1 in 85 000

Pattern of inheritance

Recessive, caused by a single-gene. An affected parent has a 25% chance of having an affected child, but only if both parents are carriers. Carriers show no symptoms.

Symptoms

Babies start with no or few symptoms, but between the ages of five and ten progressive mental deterioration occurs and children become hyperactive and disruptive. Eventually movement and speech are lost and death occurs in the mid teens.

Treatment

At present there is no cure for Sanfilippo syndrome.

Possibility of genetic testing

Sanfilippo syndrome is so rare that parents tend to find that they are carriers only when they have a child who is affected. However, for subsequent children, they can opt to have chorionic villus sampling during pregnancy to see if the foetus is affected.

Fact sheet - Thalassaemia



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Population frequency

Thalassaemia is more common in Mediterranean cultures and in those of Italian or Greek descent. It is also present in other areas including Africa, India and to a lesser extent Southeast China.

Pattern of inheritance

Recessive, caused by a single gene. An affected parent has a 25% chance of having an affected child, but only if both parents are carriers. Carriers show no symptoms.

Symptoms

Children with Thalassaemia Major appear normal at birth. As they grow they exhibit paleness and fussiness. Weakness and slow growth appear in the first or second year of life and the abdomen may swell due to an enlarged liver and spleen. The appearance of the face and head changes - the eyes appear slanted and the cheekbones become more prominent.

Treatment

Treatment for Thalassaemia involves blood transfusions every 4 to 6 weeks to sustain life. One cure can be to have a bone marrow transplant from a suitable donor.

Trials are underway for a new type of medication called deferoxamine, which can remove iron from the body. Early results have been encouraging and this promises to dramatically improve the life of Thalassaemia sufferers.

Possibility of genetic testing

A safe and reliable prenatal test has been developed to diagnose Thalassaemia in a foetus as early as 10-12 weeks after conception. Couples who are at risk may want to consider this possibility. Adults can also be tested so see if they are carriers.

Policy information sheet – Designer babies



In brief:

A term coined by the media, it is generally used to mean any case where an embryo is selected for implantation on the basis of a characteristic, for example gender or eye colour.

Detail:

The issue of designer babies has hit the headlines increasingly over the past few years. Whilst the phrase throws up images of parents choosing to have children with blond hair, blue eyes and high intelligence, the reality of the stories reported is often very different. Two particular cases have come up recently.

The Whitakers had a child in 2003 following IVF and the selection of an embryo. They were looking for a bone marrow match for their older child, who was suffering from leukaemia. The HFEA in the UK refused to allow the testing of embryos, so they went to America for their treatment. However in a similar case the Hashmis were allowed to select an embryo to be suitable as a donor for their older child. The HFEA argued that the case differed because the testing also benefited the embryo as in their case the illness was hereditary (it was passed on through their genes).

Both these cases were reported as designer babies, as they involved the selection of embryos. However, while most people would agree that selecting children to have particular traits is wrong, where it involves helping a sick sibling it becomes less clear-cut. In July 2005 the rules were relaxed to allow the selection of an embryo to be a suitable match for treating a sick sibling. However, each case will still have to be agreed by the HFEA on its own individual merits.

References:

www.hfea.gov.uk

www.bbc.co.uk/religion/ethics/issues/designer_babies

Policy information sheet – Free IVF on the NHS



The availability of IVF treatment free on the NHS varies from region to region in the UK. The NHS watchdog NICE (National Institute for Clinical Excellence) recommended to the government in early 2004 that 3 cycles of treatment should be provided on the NHS. However ministers disagreed, and have said that as of April 2005 local health authorities must provide one cycle of treatment for all women between the ages of 23 and 39.

The chance of getting pregnant with one cycle of IVF is around 25%, but this chance increases to 50% when you have three cycles. Many doctors argue that it is pointless putting a woman through the difficult process of IVF just once – it would be a much better use of resources to make three courses freely available.

Other issues arising include whether there should be an age limit for treatment, whether unmarried/gay/lesbian couples should be allowed IVF, and what to do if one or both parties already has children. Whilst for private clinics these matters are decided internally, with NHS provision the decisions can become more of a national concern.

Policy information sheet – Donor anonymity



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In brief:

Men and women who donate sperm and eggs for couples to use in infertility treatment can currently remain anonymous. From April 2005 this right will be removed.

Detail:

When a person donates sperm or eggs for use by infertile couples, their personal details are kept by the HFEA (Human Fertilisation and Embryology Authority) in a confidential database. These details cannot be released to anyone else unless the donor has given their express permission. However when children reach 18 they can ask the HFEA whether they are conceived from a donation, and if someone they intend to marry is related to them.

It was decided in January 2005 that sperm and egg donors would no longer have the right to remain anonymous. Information such as eye colour, hair colour, occupation and religion will be given on request. Donor children will also be able to go through a process to be put in touch with their genetic parents. However, as is presently the case, the genetic parents will have no legal or financial responsibility over any offspring. These changes are also not retrospective – they will not affect donations given before April 2005.

One of the strongest arguments for removing donor anonymity is that everyone has the right to know their genetic and medical history. As a person's medical notes are confidential between a patient and their doctor, this information cannot be passed onto anyone else. There is a great deal of concern that these changes will lead to a large drop in donors coming forward. Although there is no legal or financial issue, the idea of someone turning up twenty years later looking for their genetic parent is off-putting for many people. There is also almost no monetary incentive for donating in the UK – the only money donors can be paid is a small amount to cover expenses. Numbers of new donors has been dropping over recent years, and for many clinics there are now donor shortages.

Further information:

References:

<http://news.bbc.co.uk/1/hi/health/3414141.stm>

<http://news.bbc.co.uk/1/hi/health/3642437.stm>