Preimplantation Genetic Diagnosis (PGD)

Article by Alison Lashwood, Consultant Genetic Counsellor and Clinical Lead in PGD

Alison is a Consultant Genetic Counsellor and Clinical Lead in PGD. She has worked within the team at the Centre for Preimplantation Genetic Diagnosis (located in Guy’s and St Thomas’ Hospital, London) from the beginning and has helped to develop the service over the years. She is responsible for the organisation of the PGD process and sees couples in clinic to discuss the procedure and what is involved so that they can make a decision about whether they wish to proceed.

Background

Preimplantation Genetic Diagnosis (PGD) has now been available for over 20 years. The Centre for PGD at Guy’s and St Thomas’ Hospital has been offering a clinical service since 1997 and has now performed over 1000 cycles of PGD for many genetic and chromosomal disorders. PGD is now available for Spinal Muscular Atrophy (SMA) and, since 2010, Spinal Muscular Atrophy with Respiratory Distress (SMARD).

The PGD Centre is made up of health professionals working in both Genetics & Assisted Reproduction specialties, who work together to offer a comprehensive clinical service.

Options

The first time a couple find out that they are carriers of SMA or SMARD is usually following the birth of an affected child. This is a very difficult and sad time and not only often leads to the loss of their baby, but also means that the recurrence risk of 1 in 4 is something that has to be considered. Many couples would like to have further children and consider the various options available. The decision about which option to choose is a very personal one for a couple and the health professionals involved in their care will support any decision they make. The options available include:

1. Accepting the risk and taking a chance. There are 3 in 4 chances of having an unaffected baby. Testing could be offered shortly after birth to confirm whether or not a baby is affected.
2. Prenatal Diagnosis (PND) which involves testing during pregnancy from 12 weeks by chorionic villus sampling (CVS) or amniocentesis at 16 weeks, enables a couple to find out during the pregnancy whether or not their baby is affected. Many couples who have this test do so because they would not continue with an affected pregnancy. This is a very personal decision.

3. Sometimes, couples will consider egg or sperm donation. If the donor is tested to check that s/he is not a carrier of SMA/SMARD then this would avoid an affected pregnancy. This is not acceptable to all couples and it may be difficult to find a donor, however it is an option that some couples may consider.

4. PGD may be an alternative option as it helps a couple to have their own biological healthy children, but avoids termination of an affected pregnancy.

**Referrals**

We accept referrals from Regional Genetics Centres around the UK. In time we may be able to offer PGD for couples from overseas, but this is not currently possible. We do have a few eligibility criteria and these have been set as they link to the success of treatment. Couples can be seen if:

- The female partner is under 39 years at referral
- The female partner has a body mass index (BMI) of less than 30 and over 19
- Blood/DNA samples can be obtained from a couple and their affected child (stored DNA samples in local genetics centres are usually sufficient)

**Funding**

After meeting with a couple to discuss PGD the first thing we do to prepare for treatment is to apply for funding. PGD costs £8000 a cycle and we ask a couple’s Clinical Commissioning Group (CCG) to pay for this. In the past we have had over an 80% success rate on funding, but this may change over the coming years. Couples who already have unaffected children are unlikely to obtain funding. In some areas, couples who smoke will also be excluded. Unfortunately it can take many months to obtain an answer about funding and this is something over which we have no control.

If couples do not receive NHS funding, if possible, they can “self fund” which means paying the full price of £8000 per cycle.
Success

We have now treated over 900 couples with over 1000 cycles of PGD. 27 babies have been born unaffected by SMA through PGD, to couples who have previously had a child affected by SMA. The chance of having a baby following a treatment cycle improves the further through the treatment a couple progresses. The success rates are as follows:

- Per cycle started 22% (1 in 5 chance of having a baby)
- Per embryo transfer 38% (better than 1 in 3 chance of having a baby)

We now have nearly 270 PGD babies born in our centre and hope to celebrate our 300th in the early part of 2011.

Sadly not everyone who starts a treatment cycle will have embryos to transfer and some couples will miscarry after a positive pregnancy test.

If a couple has more unaffected embryos than can be transferred into the womb in a cycle, we will freeze these embryos if they are of good quality. This means that if another cycle is needed/wanted, we can offer a cycle using a frozen embryo which is far less complex than a full PGD cycle.

What is PGD?

For full details of the treatment cycle please refer to our booklet on our PGD website (see details at end of article)

A cycle of PGD is 9 weeks long and is very similar to what is offered to couples who require fertility treatment. We use medication to create a number of eggs in the ovaries and then collect the eggs under deep sedation. The eggs are then fertilised using the male partner’s sperm and the resulting embryos grow for about 3 days. The embryos then have 1 or 2 cells removed as the following diagram shows.
Stages of PGD cycle

1. Drug treatment to produce eggs
2. Scan of ovary showing development of egg sacs
3. Egg collection

4. Fertilisation of egg with sperm
5. Embryo development after 3 days

6. Embryo biopsy

7. Embryo testing
8. Embryo transfer
The cells are then tested by the molecular scientists and we can then establish which embryos are affected or unaffected. Usually 1 embryo is placed in a narrow catheter (tube) into the womb. A pregnancy test 12 days later confirms whether there is a pregnancy.

If treatment is successful then PGD pregnancies usually proceed in the same way as any other pregnancy. Special arrangements are made to look after women with twin pregnancies, although most of our pregnancies are singletons. We offer paediatric follow up for all babies born following PGD at birth, 1 and 2 years.

PGD does offer an alternative option to some couples, but it does take time to set up and unfortunately has limited success. Unfortunately it is not a suitable treatment for everyone and we do our best to advise couples as comprehensively as possible. However, it does work for some couples and if couples would like to consider the treatment further then we recommend looking at our website or asking their local Genetics Centre for advice.

After referral we can usually see couples within 6-8 weeks for an initial appointment. Then the preparation including applying for funding, setting the test up in the laboratory and organising the medical care for the couple’s treatment will take about 12 months. For further information interested couples are welcome to go to our website or telephone us for further information.

**PGD Centre website:**  [www.pgd.org.uk](http://www.pgd.org.uk)

**PGD Genetic Counsellors:**  
Tel: 020 7188 1364  
Email: [PGDGenetics@gstt.nhs.uk](mailto:PGDGenetics@gstt.nhs.uk)

Please also see our other Information Sheets:

- Chorionic Villus Sampling (CVS)
- Spinal Muscular Atrophy – Information for Families
- Who’s Who of Professionals
- Glossary of Terms
- Spinal Muscular Atrophy Support UK: Information and Support
- The Outreach Service

These can be downloaded from our website [www.smasupport.org.uk](http://www.smasupport.org.uk) or hard copies can be requested by phoning or e-mailing the support services team on: 01789 267 520  
[Supportservices@smasupportuk.org.uk](mailto:Supportservices@smasupportuk.org.uk)

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