Pre-implantation Genetic Diagnosis for Cystic Fibrosis
Edinburgh Fertility & Reproductive Endocrine Centre
Patient Information Sheet

PGD molecular testing for Cystic Fibrosis

This information sheet should be read in conjunction with the following additional information sheets:
- Preimplantation Genetic Diagnosis
- Assisted Conception Programme prospective patients information sheet

All of our genes come in pairs and we inherit one of each pair from each parent. Cystic fibrosis (CF) is caused by mutations in the CFTR gene, on chromosome 7 and the most common mutation is called delta F508, which is a deletion of the gene. A child who is affected with CF has two altered copies of the gene (and no normal functioning copies). The parents will both be carriers of CF and have one normal copy and one altered copy of the gene.

In some cases, couples are referred for PGD where one partner is affected with cystic fibrosis (with two altered copies of the gene) and their partner is a carrier of CF (with one altered copy of the gene). These couples have a higher chance of having an affected child.

In order to find out if your embryos have the faulty genes (or “mutations”) that cause Cystic Fibrosis, a cell will be removed from each one for genetic testing. The genetic material inside the cell will then be copied millions of times using a complex biochemical technique. Next, the area around the CF gene will be copied further, isolated and then examined in great detail, to identify CF mutations. This involves analysing genetic markers from around the CF gene in a similar manner to that used for “DNA fingerprinting”. The sample isolated from the embryo will be compared with samples taken from each parent, to see whether the embryo has inherited both altered copies of the gene and is therefore affected with CF. Embryos that have two functioning CF genes (unaffected) or one functioning and one altered gene (carriers) will be considered for replacement. With PGD for any condition, there is a small risk of misdiagnosis. For CF, this risk is between 0.1-2% per embryo. For this reason confirmatory prenatal testing would be offered, either by CVS or amniocentesis, as described above. PGD is limited to testing the embryos for CF. Testing for other conditions such as Down syndrome at the same time is not possible.