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## Cystic Fibrosis Screening

### What is Cystic Fibrosis?

Cystic fibrosis (CF) is a common severe disease that affects the lungs, the pancreas and the sweat glands. The gene involved in CF regulates the production of a protein in cells that controls the movement of salt in and out of cells. Symptoms of CF include repeated chest infections, chronic coughs, poor food digestion resulting in poor nutrition, and salty sweat. Cystic fibrosis sufferers have an average life expectancy of only 40 years. Currently there is no cure for CF although modern treatments enable patients to lead relatively normal lives for a time.

### What is a Cystic Fibrosis Carrier?

For a child to have cystic fibrosis both parents must be carriers of the gene mutation responsible for cystic fibrosis but they may not have CF themselves. Every cell in a person's body has two copies of Chromosome 7, one of which they inherit from each parent. If one of those copies has the mutation responsible for CF then the person is a carrier for the condition. If they have the CF mutation in both copies of Chromosome 7 then the person has CF.

### Risks of Having Cystic Fibrosis

Cystic fibrosis is the most common life threatening genetic condition affecting Australian newborns. About 1 in 25 Australians of Caucasian descent are carriers of CF. It is rare in people of Asian descent. One in every 2,500 babies, male or female, born in Australia will have CF.

### Diagnosis of Cystic Fibrosis

All newborn babies in Australia are screened for CF within a few days of birth as part of a national programme. Tests can also be done during pregnancy to determine whether the baby has CF but this is usually done only if both partners are carriers. The move now is towards a programme of prevention and testing parents prior to becoming pregnant. Couples becoming pregnant through IVF and associated procedures are uniquely placed to have their embryos screened for CF if they are carriers. This is achieved by the procedure of Pre-Implantation Embryo Genetic Diagnosis (PGD) performed on the embryos prior to embryo transfer to identify those embryos not carrying the gene mutation.

To aid this programme of disease identification and prevention, **Queensland Fertility Group** has led the way by providing Cystic Fibrosis Carrier Screening for all couples registering for infertility treatment. The costs are covered by the registration fee. The test is initially performed on the female partner. Should she be found to be a carrier then her male partner will also be tested.