CYSTIC FIBROSIS Community Care
Incorporating CFV and CFNSW

Position Statement: Cystic Fibrosis Carrier Screening

Cystic Fibrosis (CF) is the most common life threatening inherited condition in Australia. One in every 2,500 babies born in Australia each year will have CF. CF changes the way salt is transferred in and out of cells resulting in thick sticky mucus to build up in the organs causing difficulty breathing, lung damage, and impaired digestion.

There are over 2,000 gene changes (mutations) that can cause CF. One in twenty-five people on mainland Australia, and one in twenty in Tasmania are carriers of a CF gene change although most do not know this because carriers are healthy and do not display symptoms of CF. If both parents are carriers of a CF gene then there is a 25% chance of each pregnancy that the child will be born with CF. Most (> 95 %) children with CF are born to parents who did not know they were carriers, and they only find out *after* the birth of a child with CF.

Carrier screening for cystic fibrosis

CF carrier screening is a genetic test that can inform individuals if they are a carrier of the more common change in the gene that can cause CF.

Carrier screening for CF gene changes offers prospective parents the choice to find out if they are carriers of CF. It does not entirely rule out the possibility of having a child with CF, where the parents are carriers of one of the mutations that are not routinely screened for, although this is unlikely. Most CF Screening will capture just over 85% of carriers.

Carrier screening does not direct or imply a particular course of action. Understanding the risks is an important step in making an informed reproductive choice in relation to CF.

Genetic counsellors are trained to assist people to make their own choices regarding genetic testing and genetic risks. Genetic counsellors are available at most clinical genetic services.

GPs, obstetricians and Cystic Fibrosis associations throughout Australia can also provide information about CF and carrier screening to help inform people's decisions.

Cystic Fibrosis Community Care (CFCC) Position Statement on carrier screening

> *CFCC supports individuals having an opportunity to make informed life and reproductive choices as a result of CF carrier screening.*

CFCC encourages people to make an informed choice about knowing their CF carrier status as early as possible. The earlier carrier status is known the longer carriers have to make a thorough, informed and considered choice about their reproductive options including, but not limited to, understanding the impact of having a child with CF, understanding the risks of having a child with CF and exploring IVF options.

CFCC encourages people interested in understanding their risks of being a carrier for CF gene changes to consult a genetic counsellor. If a genetic counselling service is not available, CFCC encourages people to seek unbiased information to inform their personal decision making about the risks of being a carrier and of having a child with CF.

CFCC supports efforts to make carrier screening accessible for anyone who is interested in learning about their CF carrier status. This includes raising awareness of CF, raising awareness of carrier screening services and addressing other potential barriers to access and informed choice (e.g. cost of screening).