



Gene screening tests

- ◆ *Fragile X syndrome*
- ◆ *Cystic fibrosis*
- ◆ *Spinal muscular atrophy*
- ◆ *Alef8*

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Carriers About one in twenty people have faulty genes which increases their chance of having a child with a serious disorder.

Inheritance For most disorders a child is affected if both parents are carriers and both pass on their faulty gene. For others it is sufficient for the mother to be a carrier but the fault worsens when it is passed on.

Family history Despite the high risk to carriers, most affected children are born into families with no history of the disorder.

DISORDERS

Fragile X syndrome The most common cause of severe learning disability after Down's syndrome. Intellectual impairment in boys is mild to severe; girls can be normal but a large proportion are impaired. Only mothers need to be tested. *Carrier frequency 1 in 150.*

Cystic fibrosis (CF) A serious condition in which glands produce excessive amounts of thick mucus in the lungs and digestive tract. Treatment has improved but average life expectancy is still only 30-40 years. *Carrier frequency 1 in 25.*

Spinal muscular atrophy A debilitating disorder that destroys the nerve cells for walking, head and neck control, swallowing and breathing. It primarily affects children and is the main genetic cause of death in infancy. *Carrier frequency 1 in 60.*

Alef8 Seven disorders that are relatively common in Ashkenazi Jews, plus CF, in a single blood sample. *Carrier frequencies 1 in 30-100.*