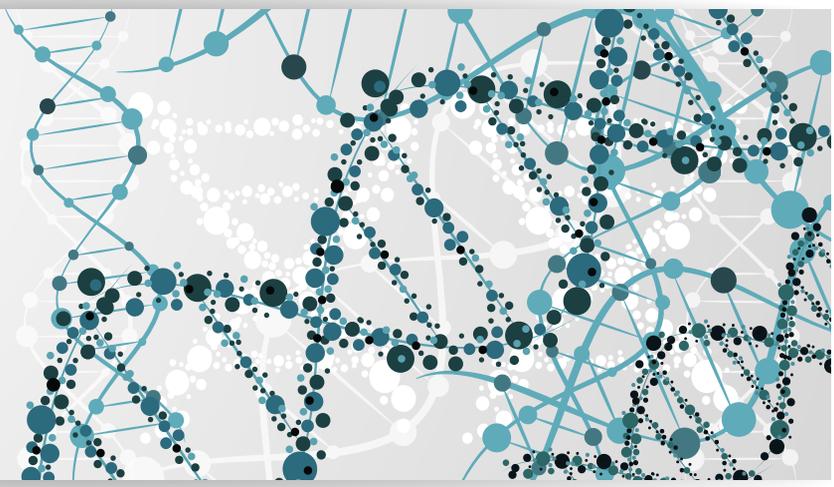


GENE Seeker

You know the risk of your child being a carrier of a genetic disease.



Smith-Lemli-Opitz syndrome

junogenetics.com

What is Smith-Lemli-Opitz syndrome?

A rare, genetic pulmonary disorder characterized by sweat, thick mucus secretions causing multisystem disease, chronic infections of the lungs, bulky diarrhea and short stature.

CF is chronic and usually progressive. Symptoms often start at birth and involve the lungs and gastrointestinal tract. A common presentation might include thick secretions and chronic infections in the lung, bulky diarrhea and short stature. Abnormal airway secretions, inflammation and infections lead to bronchiectasis and early death. CF-related diabetes (CFRD) occurs at high frequency, rising to nearly 50% of patients surviving to age 50. Male sterility is common. Individuals with mild phenotypes may have mild or absent respiratory symptoms in childhood, but some may have infertility or may develop bronchiectasis or pancreatitis later in life. These individuals are typically diagnosed by newborn screening, but may be diagnosed later in life.

What is the next step if I'm a carrier of CSmith-Lemli-Opitz syndrome?

If you are found to be a carrier of Smith-Lemli-Opitz syndrome, it is important that your partner be tested for the same genetic disorder.

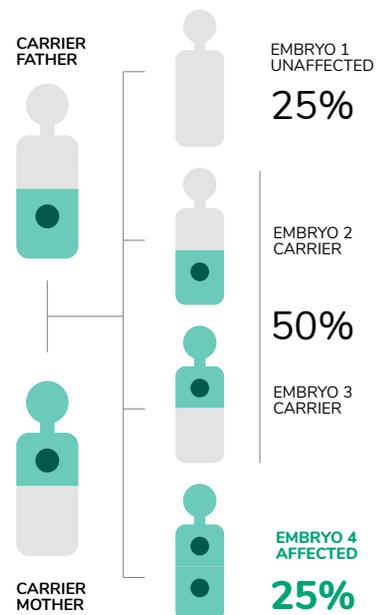
What if my partner is not a carrier?

If your partner's test for Smith-Lemli-Opitz syndrome is negative, the chance to have an affected child is low. However there is currently no test able to detect all existing mutations, so there is always a residual risk that the person who has done the test is a carrier of other less frequent mutations.

What if both me and my partner are carriers of Smith-Lemli-Opitz syndrome?

When both parents are carriers of Smith-Lemli-Opitz syndrome, the probability of having a child with Smith-Lemli-Opitz syndrome is 25%.

We recommend that you discuss your results with your doctor or genetic counselor in order to know more about reproductive options.



If both you and your partner are carriers, speak with your doctor or genetic counselor about reproductive options.

