



Genetic Information on Cleft Lip and/or Palate

General information

This leaflet has been designed to provide information about the genetic basis of cleft lip and palate.

Cleft Lip and Palate

What is an orofacial cleft?

An orofacial cleft is when a baby is born with one of the following

- cleft lip,
- cleft lip with a cleft of the palate
- cleft palate on its own.

The cleft lip can occur on one side (unilateral) or on both sides (bilateral).

Orofacial clefting can be separated into two main types:

- (1) cleft lip with or without cleft palate
and
- (2) cleft palate only .

Within babies born with a cleft palate

- 50% are cleft lip and palate,
- 30-35% are cleft palate,
- 15-20% are cleft lip only.

The genetic tendency for cleft lip, with or without cleft palate, appears to be different from the genetic tendency to cleft palate alone and it is uncommon for both disorders to occur in the same family.

In most children (70%), a cleft occurs in isolation, and the baby has no other medical problems. In the mildest form, the lip alone is unilaterally cleft, whereas in the most severe form, the lip and palate are affected on both sides.

Approximately 30% of all babies with a cleft will have other congenital conditions. For this reason, it is important that each baby will have a detailed examination to rule out any other possible conditions they may have.

Why has my child been born with a cleft?

In most children, no single cause for the cleft can be found. It is thought that most clefts are caused by a combination of many different genetic and environmental factors.

In a few families there is a strong inherited tendency for a cleft to occur. A small number of babies are born with a cleft lip and/or palate because they have a chromosome abnormality. As a rule, a child with a chromosome problem will have other health problems in addition to the cleft.

In other children, the chromosomes are normal but the child has a spelling error in the DNA (sometimes called a mutation) which alters a gene and this results in a cleft.

A “mutation” cannot be picked up by a chromosome test; one needs a genetic test of the correct gene. As each of us has 23,000 genes, it is not easy to pick the right gene to test! Routine genetic testing is not carried out in a child with an isolated cleft.

How common is it for a baby to be born with a cleft lip?

If all cleft types are counted in newborn babies, it is estimated that 1 in every 700 babies is born with an orofacial cleft. Cleft lip with or without cleft palate is most frequent in boys.

Cleft palate alone is found more often in girls.

Are there risk factors that can cause a cleft?

There is a known risk in taking certain types of drugs during pregnancy such as phenytoin, sodium valproate, benzodiazepines and corticosteroids.

There are a number of other factors that can increase the risk:

- Insulin dependent diabetes in the mother
- Maternal smoking
- Excess alcohol

Is there anything you can take to help prevent a cleft?

There is debate on the role of folic acid in the prevention of a cleft. We recommend that **every woman** thinking about becoming pregnant should take folic acid as it does help prevent other birth defects such as neural tube defects. Folic acid should be taken for 3 months before pregnancy and for the first 12 weeks of pregnancy.

If you are taking prescribed medication, we would recommend that you discuss the possible side effects with you doctor.



When in pregnancy does the cleft occur?

The lip has usually formed by 5-6 weeks of intrauterine life. The palate has formed by 10 weeks. The genes that control the formation of the lip only overlap slightly with those that form the palate.

Therefore geneticists tend to regard cleft lip and or palate as a different disorder to cleft palate on its own.

A cleft lip may be picked up before birth by a high resolution ultrasound pregnancy scan at 20 weeks or later in pregnancy. However, it is very difficult to identify a cleft palate on an ultrasound scan before birth, and most cleft palates are diagnosed after birth.

What are genes and chromosomes and why are they important?

Genes can be thought of as the 'blueprint' for our body. Each person has around 23,000 genes; these genes produce proteins that make us what we are.

Genes are so small that they cannot be seen even under a powerful microscope. They are packaged inside larger structures called chromosomes. Unlike genes, chromosomes can be seen under a microscope.

Every cell in our bodies contains a set of 46 chromosomes. They come in pairs. One of each pair comes from the mother and the other from the father. This is how we inherit characteristics from our parents. If a small piece of a chromosome is missing or there is an extra piece of any of the chromosomes, this is likely to cause birth defects and/or learning difficulties. Birth defects can also be caused by a small DNA error in a gene. This would be too subtle to be picked up on a chromosome test.

How do doctors work out the cause of the cleft in my baby?

Most babies born with a cleft have no other problems. This is known as an isolated cleft. Some babies have other birth defects and /or learning difficulties; these children are described as having a syndrome.

If the baby has more problems than just the cleft, the doctors will do further investigations such as blood tests to try and find a diagnosis, how it is likely to affect your child and if it might happen to any other children you may have in the future.

Genetic factors

The recurrence risk for a cleft for another child depends on the severity of the cleft and the presence or absence of a family history. Genetic counselling for this condition is complex. Although most are considered multifactorial disorders (genes and environmental factors).

Inheritance; Recurrence risk and offspring risk figures are available for isolated cleft lip and palate and isolated cleft palate, and take into account the severity of the malformation. If the cleft is associated with a known syndrome, the recurrence risk will depend on the genetics of the syndrome.

Genetic counselling can identify high-risk families. The cleft team will refer those families where they think the child has a genetic syndrome. Talk to your cleft team about accessing the genetics service if you are worried.

The recurrence risk is increased if the cleft is more severe.
If you would like to access specialist information on cleft lip and palate please go to:-

CLAPAI Cleft Lip & Palate Association of Ireland
www.clapa.ie

Affected Person in Family	Risk of Another Affected Child
*Sibling with cleft lip on one side	2- 3%
*Sibling with cleft lip and palate on one side	4%
*Sibling with cleft lip and palate both sides	5-6%
Two affected siblings	10%
*Affected sibling and parent	10%
Affected parent	4%

*Brother(s) or Sister(s) risk to the next child in the family



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The information contained in this leaflet is correct at time of print