What is Cri-du-Chat syndrome?

The name of this syndrome is French for "cry of the cat," referring to the distinctive cry of children with this disorder. The cry is caused by abnormal larynx development, one of the many symptoms associated with this disorder. It usually becomes less noticeable as the baby gets older, making it difficult for doctors to diagnose cri-du-chat after age two. Cri-du-chat is caused by a deletion (the length of which may vary) on the short arm of chromosome 5. Multiple genes are missing as a result of this deletion, and each may contribute to the symptoms of the disorder. One of the deleted genes known to be involved is TERT (telomerase reverse transcriptase). This gene is important during cell division because it helps to keep the tips of chromosomes (telomeres) in tact.

How do people get Cri-du-Chat syndrome?

A deletion is caused by a break in the DNA molecule that makes up a chromosome. In most cases, the chromosome break occurs while the sperm or egg cell (the male or female gamete) is developing. When this gamete is fertilized, the child will develop cri-du-chat syndrome. The parent, however, does not have the break in any other cells of the body and does not have the syndrome. In fact, the break is usually such a rare event that it is very unlikely to happen again if the parent has another child.

It is possible for a child to inherit a broken chromosome from a parent who also had the disorder.

What are the symptoms of Cri-du-Chat syndrome?

Babies with cri-du-chat are usually small at birth, and may have respiratory problems. Often, the larynx doesn't develop correctly, which causes the signature cat-like cry.

People who have cri-du-chat have very distinctive features. They may have a small head (microcephaly), an unusually round face, a small chin, widely set eyes, folds of skin over their eyes, and a small bridge of the nose.

Several problems occur inside the body, as well. A small number of children have heart defects, muscular or skeletal problems, hearing or sight problems, or poor muscle tone. As they grow,
people with cri-du-chat usually have difficulty walking and talking correctly. They may have behavior problems (such as hyperactivity or aggression), and severe mental retardation. If no major organ defects or other critical medical conditions exist, life expectancy is normal.

**How do doctors diagnose cri-du-chat syndrome?**

Doctors most often identify cri-du-chat by the infant's cat-like cry. Other signs are microcephaly, poor muscle tone, and mental retardation.

It is also possible to test for cri-du-chat (and other chromosomal abnormalities) while the baby is still in its mother's womb. They can either test a tiny sample of tissue from outside the sac where the baby develops (chorionic villus sampling (CVS)), or they can test a sample of the amniotic fluid (amniocentesis).

**How is cri-du-chat syndrome treated?**

Although there is no real treatment for cri-du-chat syndrome, children with the disorder can go through therapy to improve their language skills, motor skills, and to help them develop as normally as possible.

**Interesting facts about cri-du-chat syndrome**

The geneticist Jerome Lejeune identified cri-du-chat syndrome in 1963. He also discovered the genetic abnormality that causes Down syndrome.

Cri-du-chat is one of the most common syndromes caused by a chromosomal deletion. It affects between 1 in 20,000 and 1 in 50,000 babies. In 80 percent of the cases, the chromosome carrying the deletion comes from the father's sperm rather than the mother's egg.

When deletions occur during the formation of an egg or sperm, it is caused by unequal recombination during meiosis. Recombination normally occurs between pairs of chromosomes during meiosis while they are lined up at the metaphase plate. If the pairs of chromosomes don't line up correctly, or if the chromosome breaks aren't repaired properly, the structure of the chromosome can be altered. When unequal recombination occurs at this location on chromosome 5, it causes cri-du-chat syndrome.