Genetics in Cri du Chat Syndrome

It is important to remember that a person is a person first, and the diagnosis is secondary

Prior to reading about the genetics of Cri du Chat syndrome (CdC), if you would like to read more about the basic concepts of genetics go to our key topics area and select 'genetics' from the drop down menu.

Cri du Chat syndrome is caused by a missing piece of information (deletion) on the short arm of chromosome 5, and is also known as 5p minus syndrome.

The size of the deletion can vary. There is a 'critical region' on chromosome 5 that appears to be specifically related to the characteristic features of Cri du Chat syndrome. This is the region called '5p15.2'. In addition, the region called '5p.15.3' appears to be related to specific features of Cri du Chat syndrome, for example, the cat-like cry and speech delay have been mapped to 5p15.3.

Is Cri du Chat syndrome an inherited condition?

In 85% of individuals the chromosomal deletion that causes Cri du Chat syndrome occurs at random, therefore, it is not inherited from either parent. This is called a de novo deletion. In 10-15% of individuals the syndrome is inherited from a parent who carries a rearrangement involving the short arm of chromosome 5 (familial deletion). It is estimated that 90% of familial cases are due to parts of the parental chromosome being swapped around (balanced chromosomal translocation). It is estimated that 5% of cases are due to an inversion of 5p. This is when part of 5p (the small arm of chromosome 5) breaks off and get reinserted and reattached in its original position but in an inverted (upside down) position.

More information on genetic testing can be found at:

NHS - Genetics & Genetics Testing