# Diagnosis of Cri du Chat Syndrome

## A high pitched vocal sound is usually the first sign that a child may have Cri du Chat syndrome.

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**Clinical Diagnosis**

Cri du Chat syndrome can be diagnosed clinically, by observing distinctive facial and other physical characteristics that are common in this condition.

A clinical diagnosis can be given by a paediatrician, GP or a clinical geneticist, they will look at the physical and behavioural characteristics of a child and from this information determine whether the child has the syndrome.

**Genetic Testing**

Diagnosis may be made by genetic testing. Someone with a genetic diagnosis will have had a genetic test. A genetic test involves taking a sample of blood or saliva and sending it to be tested for the Cri du Chat syndrome deletion on chromosome 5.

Individuals with typical Cri du Chat syndrome (where the genetic deletions are within the critical region - 5p15.2) are the most affected, and have severe to moderate intellectual disabilities.

Individuals with atypical Cri du Chat syndrome (those with genetic deletions outside the critical region of 5p15.2) often have much milder intellectual disabilities, and subtler facial and behavioural features then those with typical Cri du Chat syndrome.

For more information about diagnosis, visit the Cri du Chat support group page

To read more about some of the genetic tests used for diagnosis click here.