Physical Characteristics

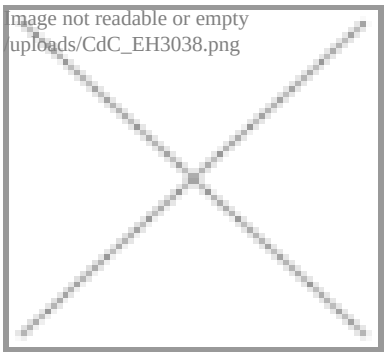
Research into Cri du Chat syndrome has shown that the larger the size of the chromosomal deletion, the more pronounced the physical characteristics are among individuals with Cri du Chat syndrome.

Some people believe that the descriptions of physical characteristics of a syndrome are not helpful to individuals; however, we believe that knowing the physical characteristics is important. To read about why we have included physical characteristics on this website go to the key topics area and select 'physical characteristics' from the drop down menu.

An individual with Cri du Chat syndrome may have many of the following physical characteristics or only a select few. Please note that the physical characteristics of Cri du Chat syndrome are very broad and not all individuals with Cri du Chat syndrome will have all of facial characteristics listed below:

Facial Characteristics

The most common facial characteristics are:



Emily

- Rounded face
- Widely spaced eyes
- An extra fold of skin at the inner corners of the eyes (epicanthal folds)
- Flattened and widened nasal bridge
- Ears that are positioned low on the head
- Microcephaly (small head size)
- A short neck

Studies show that facial characteristics change over time, in particular, lengthening of the face and coarsening of features.

Physical Characteristics

The distinctive cat-cry is a core feature of the syndrome. The cry is thought to be caused by anomalies in the larynx (voice box), which is smaller and diamond shaped, and anomalies to the cartilage located in the throat behind the tongue and in front of the larynx, which is usually smaller and more floppy than in typical infants.

Poor muscle tone (hypotonia) may initially cause a poor sucking response in new-borns with Cri du Chat syndrome. It can also contribute towards coordination difficulties as the child gets older.

Many infants tend to have low weight and this usually persists for the first two years of life. Gastroesophagael reflux (similiar to heart burn) is also common during the first years of life. To read more about health in Cri du Chat syndrome click here.

Although some individuals with Cri du Chat syndrome may experience serious medical conditions, most individuals will have a typical life expectancy.