

Health Issues in Cri du Chat Syndrome

Parents and carers have told us that being presented with a long list of health difficulties can be daunting. However, they also reported that having this list available can be reassuring and can help them to watch out for early warning signs. The key message here is that children with Cri du Chat syndrome may be prone to some health difficulties; however, with appropriate support and regular health checks with an informed professional, a lot can be done to reduce the impact on an individual's well-being.

Vision and Hearing

Most individuals with Cri du Chat syndrome do not have difficulties with vision. When visual problems do occur they include near sightedness, wobbly eyes, and optic nerve abnormalities

Hearing is a relative strength for individuals with Cri du Chat syndrome because difficulties hearing are uncommon. However, between 70-80% of people with Cri du Chat syndrome show hypersensitivity to noise (hyperacusis). While hearing is a general strength, it is important to note that middle ear infections (otitis media) are common in individuals with Cri du Chat syndrome and this can affect hearing.

Mobility

Individuals with Cri du Chat syndrome will have some degree of mobility, and many are able to walk, although almost all will experienceco-ordination, balance and motor-control difficulties.

In 2006, a study of 91 children with Cri du Chat syndrome found that by three years of age half the children could walk by themselves and all of the participants in this sample walked later, however, development of mobility was delayed.

Feeding

Many infants with Cri du Chat syndrome have feeding difficulties in their first year, such as a poor sucking reflex, vomiting and gastroesophageal reflux. In one study, 63% of parents reported feeding difficulties during their child's neonatal period.

Often difficulties with sucking, chewing and swallowing lead to a delayed introduction to solid foods. In one study, 37% of children and adolescents with Cri du Chat syndrome always ate food which had been pureed. Around 21% had a typical diet and the rest had a combination of typical foods with some being pureed. This finding could be age related and suggests that there may be a delayed progression onto normal foods, from pureed foods.

Dribbling

Many children with Cri du Chat syndrome will experienceexcessive dribbling. This may be due to weak muscle control and may be helped by speech therapy, medications or surgery to relocate the saliva glands towards the back of the mouth under the tongue. This helps the saliva to go down the throat and be swallowed.

Reflux and Respiratory Tract Infections

We do not yet know how often problems with gastro-oesophageal reflux occur in young children and adults with Cri du Chat syndrome but it may be helpful to be aware that these difficulties can occur.

Many individuals are prone to recurrent upper respiratory tract infections and dental problems. In other individuals with intellectual disability, proneness to respiratory tract infections and dental problems appear to be linked to the presence of reflux (severe heartburn). Sometimes, pain and discomfort related to gastro-intestinal or dental complications may be associated with <u>self-injurious or behaviours that challenge</u>.

Curvature of the Spine

Many individuals with Cri du Chat syndrome are prone to developing a curvature of the spine (scollosis). This can become more apparent with advancing age. This should be regularly monitored in all children and adults with Cri du Chat syndrome, particularly during puberty when children have a growth spurt.

Constipation

It is reported that around 7 out of 10 children and young adults have significant problems with constipation, and a high-fibre diet may help with this.

It is always important to ensure that any behaviours that challenge observed are not <u>underpinned by pain</u>.

For more information on the signs of pain in children with severe intellectual disability and/or communication difficulties please visit the <u>Cerebra webpage</u>.

Peripheral Sensory Neuropathy

There is some evidence that people who have Cri du Chat syndrome might have a peripheral sensory neuropathy.

This disorder can have two important effects that might be relevant to self-injury. First, pain may not be experienced in the 'normal' way; painful stimuli may not be experienced as painful. Secondly, people may

experience unusual sensations in their hands and arms. These sensations are described by some people as pins and needles (dysaesthesia) or a mild burning sensation. It has been suggested that peripheral sensory neuropathy may be associated with behaviours that challenge underpinned by pain.