Health Issues in Cornelia de Lange Syndrome

Eye problems:
Eye problems associated with Cornelia de Lange syndrome include short sightedness, blocking of the tear duct which is often associated with infection and inflammation of the lining of the eye (conjunctivitis) and the eyelids (blepharitis). An inability to fully open the eyes (ptosis) is common and can be a major problem.

Ear problems:
Commonly reported ear problem in Cornelia de Lange syndrome are chronic otitis media (middle ear infection) and mild-to-moderate hearing loss (in the absence of otitis media).

Nasal issues:
Sinusitis (infection of the sinuses) is a common problem in individuals with Cornelia de Lange syndrome which may be associated with nasal polyps. Inflammation of the sinuses in the nose causes pain and discomfort to the individual. Cleft palate occurs in 20% of cases and high arched palate is very common. Inflammation of the ear canal (otitis externa) is seen reasonably frequently.

Teeth:
Proper dental care is important for everyone and is recommended even during infancy.

Parents and carers of people with Cornelia de Lange syndrome should be aware of the following problems:

- Small jaw development
- The erosion of teeth caused by stomach acids from reflux
- Thin enamel.
- Crowded teeth
- Extra or missing teeth
- Poor oral hygiene due to the difficulties in teeth cleaning.
- Cavities
- Periodontal (gum) disease
- Small teeth
- Teeth grinding

Among the great number of dental problems common for people with Cornelia de Lange syndrome, the most serious may be those relating to reflux. Reflux and the acid that enters the mouth can cause a serious breakdown of the enamel covering the teeth.

Once the protective layer of enamel has been altered, large dental cavities can destroy the teeth in a short period of time. Often, it is extremely difficult to provide adequate oral hygiene for the individuals due to their protective and resistant actions, inhibiting access to the mouth.

Getting a toothbrush to the back-most areas of the mouth where the acid can have its most devastating effects can be a challenge. The Cornelia de Lange Syndrome Foundation's dental experts recommend the Collis Curve toothbrush which has eased the teeth cleaning chore for many families. These brushes are available online or through the Foundation office www.cdls.org.uk

Bone and joint problems:
Scoliosis (curvature of the spine) occurs more commonly than in children without Cornelia de Lange syndrome. Restricted movement at the elbow joint is very common in Cornelia de Lange syndrome.

Abnormalities in the hip occur in more than 10% of children and adults with the syndrome. This is characterised by increasing shallowness of the normally 'cup shaped' hip socket until the often unusually shaped 'ball shaped' upper end of the thigh bone (femur) is at risk for dislocation, interfering with the person's ability to walk. This condition is difficult to detect through a physical examination and therefore it may be wise to obtain an x-ray of the hips every few years during growth.

Neurology:
Seizures are reported in 25% (or 2 - 3 out of 10) of children with Cornelia de Lange syndrome. A seizure is an abnormal discharge of electrical activity in the brain.

Seizure activity may be suspected when people with Cornelia de Lange syndrome show changes in behaviour. Because not every seizure disorder appears on an EEG (a medical procedure that monitors brain activity), descriptions of behaviour surrounding the time of observed change are very important.

Most seizure activity can be controlled by medications. However, some seizures are extremely difficult to control without significant 'trade offs' between control of the seizure and heavy medication side effects.

Peripheral sensory neuropathy.
There is some evidence that children and adults who have Cornelia de Lange Syndrome might have a peripheral sensory neuropathy. This means, that part of the nervous system that is associated with pain and sensation (peripheral nervous system) might not be sending the right signals to the brain. Given that for many children and adults the upper limbs do not develop properly it would not be surprising if the peripheral nervous system in the arms had also not developed normally. Even if the hands and arms are well-formed it is entirely possible that a peripheral sensory neuropathy is still present.

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.

Other health issues to be aware of:

Heart defects:
Around 25% of children with Cornelia de Lange Syndrome are born with a heart condition of some sort.

The most common heart problems include:

- Ventricular septal defect (VSD; a hole in the wall between the right and left ventricles of the heart).

- Atrial septal defect (ASD; a hole in the wall between the atria, the upper chambers of the heart).
- Patent ductus arteriosus (failure of the ductus, a foetal structure, to close after birth).
- Tetralogy of fallot (TAF; a combination of four heart problems that occur together from birth including VSD).

Heart disorders are usually signalled by a heart murmur. When a problem is found further evaluation by a paediatric cardiologist may be necessary. Since most of these conditions are detected in the first three months of life, caregivers of older children should not worry needlessly about the development of heart problems.

Surgery is not usually necessary for the most commonly occurring heart problems in Cornelia de Lange syndrome.

Undescended testes:
Undescended testicles are very common in males with Cornelia de Lange syndrome. This is due to failure of the testes to come through the inguinal canal that connects the abdomen and the scrotum. There are risks associated with not bringing them down, including heightened risk for developing testicular cancer, hernias, and twisting of the testicle.

It is generally advised that undescended testicles are repaired surgically as early as possible. Testicles should then be monitored after surgery to ensure that they do not go back up into the abdomen. If they just rise into the canal, but can be felt then there is no increased risk.

Raynaud's phenomenon:
Raynaud's phenomenon is a condition that affects the blood vessels in the the fingers and toes, and is characterised by episodic attacks, called vasopastic attacks, in which the blood vessels in the fingers and toes narrow, usually in response to cold temperatures and/or emotional stress. It is thought that some individuals with Cornelia de Lange Syndrome may be affected by Raynaud's phenomenon.