

Physical Characteristics of Lowe Syndrome

An individual with Lowe syndrome may have many of the following physical characteristics, or only a select few. Please note, the physical characteristics of Lowe syndrome are quite broad and not everyone will have these characteristics. However, a couple of the features are present in nearly all boys with Lowe syndrome.

Facial Characteristics

There are a number of physical characteristics regarding the eye specifically. These include:

- Cataracts are present at birth in nearly all cases of Lowe syndrome; they can make the eyes look cloudy and impair vision.
- Infantile glaucoma occurs in approximately half of individuals with Lowe syndrome- this is where there is too much pressure in the eye, and this can cause eyes to become enlarged or look ‘bulging’ (exophthalmos).
- Some children with Lowe syndrome develop crossed eyes or a squint.

With regard to head and face shape, head circumference is generally typical, but elongation of the face is sometimes a feature of Lowe syndrome.

Individuals with Lowe syndrome may have prominent foreheads, chubby cheeks, deep-set eyes, a high arched-palate and a fair complexion.

There may be a delayed eruption of ‘adult’ teeth and overcrowding of teeth in the mouth. As well as this, individuals with Lowe syndrome may have white spots on their teeth due to thin enamel and excessive calcium deposits.

Movement

Due to weak muscles, babies with Lowe syndrome may appear floppy shortly after birth; this is referred to as hypotonia. People with hypotonia may also have poor reflexes on physical examination, a weak cry and be unable to suck properly, which causes them to be underweight in their first year. This tends to improve with age but normal motor tone and strength are rarely achieved.

Decreased motor tone leads to delayed motor milestones. There may be difficulties with sitting, standing and walking because of this, but 75% of boys with Lowe syndrome are able to walk independently between the ages of 6 and 13.

Around half of individuals with Lowe syndrome develop scoliosis (curvature of the spine), and some have an abnormally rounded back.

Rickets (soft bones) used to be common in Lowe syndrome, and caused the bones to become bowed- this especially affected the legs (bowed legs). This complication can be prevented with medical treatment.

Inflammation around the tendons (tenosynovitis) and joint inflammation (arthropathy) occur in some individuals with Lowe syndrome.

Other Physical Characteristics

Individuals with Lowe syndrome have a short stature. Birth length is generally typical, but by their first birthdays individuals with Lowe syndrome have fallen below the 10th percentile for height. People with Lowe syndrome do grow, just at a slower rate, and the average adult height is 155cm (5 feet).

Puberty may be delayed in onset, but other than this, sexual development is typical. In approximately a third of boys, cryptorchidism (undescended testes) may occur.

Cysts in the mouth and skin may appear in areas such as the gums, buttocks and lower back. These can cause pain and become infected.