

# Physical Characteristics of Kleefstra Syndrome

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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.

It is important to note that children with Kleefstra syndrome vary in the number of physical characteristics that they show.

## Facial Features

- Widely spaced eyes
- Eyebrows that grow together,
- Midface hypoplasia (when the centre of the face develops more slowly than the eyes, forehead and jaw)
- Protruding tongue
- Nostrils that open to the front rather than downward
- Everted lower lip (rolled out lips)
- A large tongue
- Protruding lower jaw

## Other physical characteristics

- High birth weight (the average birth weight is 3.5 kg) and childhood obesity.
- Congenital (from birth) heart and kidney problems – a high number of individuals with Kleefstra Syndrome have congenital heart defects (around half of individuals). Kidney defects are less common (1-3 in 10 individuals).
- Hypotonia (poor muscle tone) – hypotonia means that babies appear floppy.
- Minor genital anomalies in males.
- Catatonia (particularly in rigidity of arms and repetitive movement of the fingers).

*The information on this website was developed in collaboration with [www.kleefstrasynndrome.org](http://www.kleefstrasynndrome.org). This is the parental support group for Kleefstra, a well-developed community with an active facebook group and parent support conference. Please visit their website for further information and to get in touch with families of individuals with Kleefstra syndrome.*