

Why am I so short?

A common reason for referral to the genetic counselling service is short stature. Patients often ask why they are short and if there is any treatment. To answer these questions, it is necessary to start from the basics.

An individual's final adult height depends on many factors, including ethnicity, parental height, nutritional intake and environmental factors. Furthermore, body systems (such as the circulatory and endocrine systems) have to function properly to ensure normal growth and development of the child. Short stature is defined as body height less than the 3rd centile in the same age group. If doctors find that children are short or their growth velocity is significantly slower than normal, further investigations would be initiated to identify the underlying cause. Appropriate treatment would be given if possible.

A common genetic cause of short stature is Turner Syndrome. This syndrome occurs only in females, with a frequency of one in every 2,500 women. Most cases are sporadic with no other family members affected. The commonest underlying genetic mechanism of this disorder is monosomy X, i.e. patients have only one X chromosome (constituting a karyotype of 45,X). Some patients have mosaic Turner syndrome where only a proportion of their body cells has monosomy X. Other patients with Turner variants have a structural abnormality on one of the X chromosomes.

Patients with Turner Syndrome may have characteristic features like short stature (adult average height of about 145 cm), cubitus valgus, short metacarpal and metatarsal bones, lymphoedema, webbed neck, shield-like chest and widely-spaced nipples. The majority of patients have ovarian dysgenesis, amenorrhoea and infertility. In addition, some patients have congenital heart diseases, renal anomalies or thyroid disorders.

Turner syndrome is not curable, but supportive treatments are available to alleviate some of the symptoms. These include growth hormone therapy to improve growth velocity and height, hormonal replacement therapy to promote the development of secondary sexual characteristics and to prevent osteoporosis, as well as the use of assisted reproductive technologies to increase the chance of pregnancy.

