Defective genes

Mucopolysaccharides (MPS) are long chains of sugar molecules which form an important component in connective tissues. Mucopolysaccharidosis (MPS) is a group of rare inherited metabolic disorders, in which a specific enzyme is missing or insufficient. This can lead to excess build-up of mucopolysaccharidosis in body tissues, which in turn affects a person’s normal body function.

The disorder may not be apparent at birth, but signs and symptoms develop with age as more cells get damaged. There are seven subtypes of MPS with varying effects and degrees of severity. The severity depends on the degree of enzyme deficiency and may include abnormal facial appearance, bone and cartilage deformities causing a person to be short stature, or have physical disabilities, corneal opacities, mental retardation, cardiac and respiratory abnormalities.

MPS is a genetic disorder caused by a recessive gene from each of his or her parents. A subtype called MPS II is the only exception in where a mother can pass the disease directly to her child, and only if the child is a male. A couple's chance of having another child with MPS is 1 in 4. The overall incidence of MPS is about 1 in 25,000 births.

There is no cure for MPS at present, but early diagnosis can facilitate treatment. Early haematopoietic stem cell (bone marrow or cord blood) transplantation is useful in the case of MPS I and VI. Recently, enzyme replacement therapy has also been shown to be effective in dealing with MPS I, II and VI symptoms. However, this is very expensive and involves lifelong treatment.