Newborn Metabolic Screening Program

CUHK-BCM Joint Centre for Medical Genetics
The Chinese University of Hong Kong

Information for Parents
Screening for

Mucopolysaccharidosis 1

(MPS 1)

Enquiries:
☎ (852) 5569 6412 (office hours)
☎ (852) 3505 4219 (voicemail service)

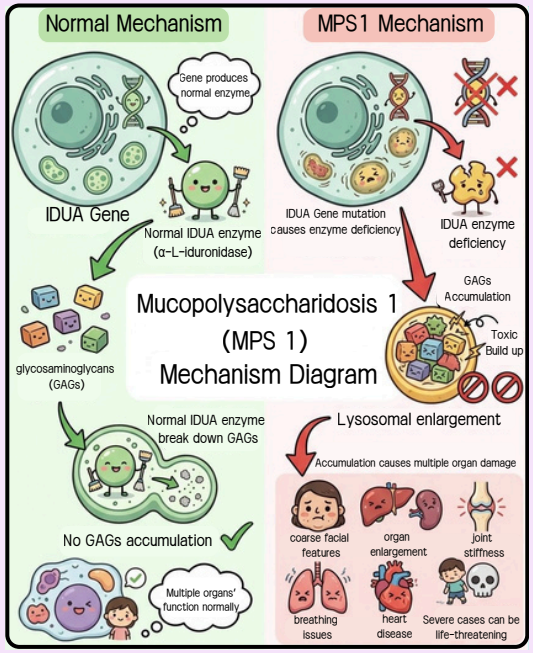
website:

If you wish to join this screening program,
please contact your obstetrician during antenatal period
or contact your paediatrician within 7 days after delivery

What is Mucopolysaccharidosis 1 (MPS 1)?

Mucopolysaccharidosis Type 1 (MPS1) is a rare inherited disorder caused by mutations in the IDUA gene. This gene produces the enzyme α-L-iduronidase, essential for breaking down glycosaminoglycans (GAGs).

Enzyme deficiency causes GAGs accumulation, disrupting multiple organ systems. Toxic buildup damages bones, joints, heart, airways, and nerves, leading to coarse facial features, organ enlargement, joint stiffness, breathing issues, heart disease, and developmental delays. Severe cases can be life-threatening.



What are the symptoms of MPS1?

MPS1 is classified into three types based on severity:

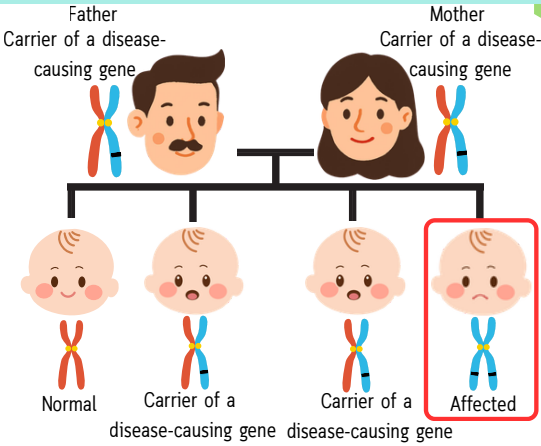
Mild (Scheie) :
Onset after age 5, normal intelligence, mainly joint, vision, and heart issues.

Intermediate (Hurler-Scheie) :
Onset at 1-5 years, may have mild intellectual impairment, primarily joint stiffness.

Severe (Hurler) :
Symptoms appear before age 1, including neurodegeneration, skeletal deformities, and heart disease. Untreated survival <10 years

Early diagnosis via newborn screening can improve outcomes.
Delayed diagnosis may lead to irreversible damage.

How is Mucopolysaccharidosis 1 (MPS 1) inherited?

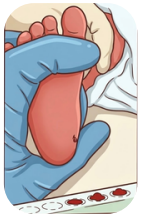


MPS 1 is an autosomal recessive condition, which means that a baby must inherit one disease-causing CFTR gene variant from each parent to be affected. If both parents are carriers, each pregnancy has 1/4 chance of having Cystic Fibrosis, a 1/2 chance of being a carrier without any symptoms and a 1/4 chance of not carrying any affected gene at all.

Mucopolysaccharidosis 1 (MPS 1) screening and its aim

The MPS 1 screening aims to detect babies at risk of MPS 1 as early treatment can prevent severe disability.

A few drops of blood are collected onto a card by pricking the baby's heel in its first 24 hours to 7 days of life, and the concentration of IDUA enzyme activity is measured.



All babies with a positive screening result will have to undergo further investigations to confirm whether they are affected by MPS1 or not.

Outcomes of MPS 1 patients

Early diagnosis is crucial for MPS 1. Enzyme Replacement Therapy (ERT) is given to manage physical symptoms, but it does not prevent cognitive decline.

For severe cases (Hurler syndrome), early hematopoietic stem cell transplantation (HSCT) is recommended to improve long-term outcomes and help preserve brain development.

A multidisciplinary team is needed for ongoing care, including physical therapy and surgical interventions. During illness, standard supportive care is advised. There are no specific dietary treatments, but good nutrition supports overall health.