Lesch-Nyhan disease

A guide to HPRT1-related disease, specialist assessment and the coordination of day-to-day care.

New editorial reference, 20 November 2026. Open the historical SSBP information sheet.

Understand the condition and its range

Lesch-Nyhan disease is a rare inherited condition caused by disease-causing changes in HPRT1. This gene provides instructions for an enzyme that helps cells recycle some of the building blocks used in DNA and RNA. When enzyme activity is reduced, the body makes too much uric acid. Uric acid can contribute to kidney stones and joint problems. The classic form also affects movement, development and behaviour.

HPRT1-related conditions form a range. Some people have uric acid complications alongside fewer problems with movement or behaviour, and some mainly have problems related to uric acid. In the classic form, low muscle tone may be noticed early and dystonia can develop. Dystonia causes involuntary muscle contractions that affect posture or movement. The pattern of symptoms can change as a child grows, and the care team can explain which concerns apply to an individual.

Arrange assessment and keep a useful record

In the UK, speak to a GP or paediatrician about concerns involving delayed movement, unusual muscle contractions or unexplained kidney stones. A specialist referral may involve paediatric metabolic medicine, neurology or clinical genetics, depending on the findings and local pathway. A short timeline can help. Include developmental milestones, urinary problems, pain episodes, previous assessments and any relevant family history.

Specialists consider the overall pattern and may use genetic or biochemical testing to establish whether someone has an HPRT1-related condition. The clinician can explain how a result relates to the person's needs and which follow-up is appropriate. HPRT1-related disease is usually inherited through the X chromosome and most often affects boys. Genetic counselling can explain why the pattern differs across families and whether relatives may want to consider testing.

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Coordinate physical and behavioural care

Self-injurious behaviour is a feature of the classic form. It can be hard for the person to control and can be distressing for them and for those who support them. Ask the team to agree an individual safety plan together with the person and caregivers. The plan can cover early signs, how the person communicates pain and which responses have helped. New or worsening episodes can be a reason to look for pain, illness or changes in routine as part of a clinical review.

Uric acid care and support for movement or behaviour are separate parts of the same plan. Ask who will monitor kidney and joint concerns and who will help with seating, mobility, communication and safe eating or drinking if needed. Physiotherapy, occupational therapy and speech and language therapy can help adapt equipment or routines in line with the person's priorities.

Plan for changes across home, school and services

Keep a short care summary that can travel between settings with the person's agreement. Include how they show discomfort, known stressors, effective communication aids, equipment and whom to contact about urinary problems, pain or injury. When several people provide care, note which clinician is responsible for each follow-up and when it will happen.

Before a major change, such as moving to another school or transferring to adult services, review the care summary together. Ask what the person wants to do more independently and which adjustments help them take part. If needs have changed since the last review, bring a concrete example and ask which team can address it.

Sources and further reading

GeneReviews, HPRT1 disorders (www.ncbi.nlm.nih.gov)

Jinnah and colleagues, HPRT1-related clinical spectrum (academic.oup.com)

NHS England Genomics Education Programme, roles in clinical genetics (www.genomicseducation.hee.nhs.uk)