Hunter Syndrome

Gene Tested: *IDS*

Also Known As:

- Mucopolysaccharidosis type II (MPS II)
- Iduronate 2-sulfatase (IDS) deficiency

Disorder: Hunter syndrome is a relatively rare lysosomal storage disease which is caused by a deficiency of iduronate-2-sulfatase (IDS). Hunter syndrome is inherited as an X-linked recessive disorder. It is estimated that about 1 in every 100,000 newborn males has Hunter syndrome.

Hunter syndrome is a progressive disorder with a highly variable phenotype. Typically, newborns with Hunter syndrome show no signs of the condition. A child with more severe Hunter syndrome may have decelerating growth during the first two years of life and developmental delay. They may develop frequent ear and sinus infections, rhinorrhea, breathing difficulties, and a gradual coarsening of facial features. The liver and spleen are typically enlarged, abdominal and/or inguinal hernias are common, and behavioral hyperactivity or irritability is often reported. Gradually, bone and joint problems develop, respiratory complications increase, and the child may begin to lose developmental skills. Unfortunately, many individuals with more severe Hunter syndrome die before reaching adulthood.

A child with milder Hunter syndrome typically does not have the developmental or behavioral concerns seen in the more severely affected children. However, mildly affected individuals may have many of the same physical characteristics seen in more severely affected individuals, although these characteristics tend to develop at a slower rate.

Problems with the bones and joints, respiratory difficulties, hearing loss, and later-onset heart disease are common medical complications of mild Hunter syndrome. Individuals with milder symptoms may have a near-normal life expectancy.

The diagnosis of Hunter syndrome can be confirmed by any one of three different methods: 1) documenting increased urinary excretion of dermatan sulfate and heparan sulfate; 2) measuring iduronate 2-sulfatase activity in skin fibroblasts, plasma, or peripheral leukocytes to determine deficiency or; 3) by documenting the presence of a mutation in the *IDS* gene. Genetic confirmation of the mutation in the proband allows for accurate carrier testing in at-risk females and prenatal diagnosis or preimplantation genetic diagnosis in future pregnancies.

Indications:

- Confirmation of diagnosis in a symptomatic individual
- Presymptomatic testing of at-risk relatives
- Carrier identification in individuals with a family history of Hunter syndrome
- Prenatal diagnosis of an at-risk fetus, after confirmation of mutation in the mother or sibling (by prior arrangement only)

Specimen:

Blood samples should be collected in EDTA (lavender topped) tubes. A minimum of 5 mLs on adult or 3 mLs on child is required for analysis. Blood collected on Friday may be stored in refrigerator until Monday for overnight shipment.

For other tissue requirements please call 1-800-344-2462, extension 4474.

Cincinnati Children's
Human Genetics

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CLIA#: 36D0656333
Phone: (513) 636-4474
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Email: moleculargenetics@cchmc.org
www.cincinnatichildrens.org/molecular-genetics

Testing Methodology:
PCR-based sequencing of all eleven exons and exon/intron boundaries of the *IDS* gene.

Test Sensitivity:
PCR-based sequencing of the *IDS* gene identifies a mutation in ~80% of male patients with Hunter syndrome. The sensitivity of DNA sequencing is over 99% for the detection of nucleotide base changes, small deletions and insertions in the regions analyzed. Multiple exon deletions and recombinations with the *IDS* pseudogene, which account for approximately 20% of *IDS* mutations, are not detected by sequence analysis. *IDS* is the only gene associated with Hunter syndrome.

Turn-Around Time:
Reports are routinely available within 21 days of sample receipt. Abnormal results will be called to the referring physician. All reports will be faxed to the referring physician.

CPT Codes:
- IDS Full Gene Sequencing 81405
- Family Mutation Studies 81403

Please call 1-866-450-4198 for pricing, insurance preauthorization, or with any billing questions.

Results:
Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Additional information and test requisitions are available at: www.cchmc.org/molecular-genetics

Shipping Instructions:
Please enclose **test requisition** with sample.
All information must be completed before sample can be processed.

Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday

Ship to:
Cytogenetics and Molecular Genetics Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474