

Iduronate-2-Sulfatase, Blood Spot

Patient ID SA00060930	Patient Name SAMPLEREPORT, I2SBS A		Birth Date 1976-06-10	Gender F	Age 37
Order Number SA00060930	Client Order Number SA00060930	Ordering Physician Client, Client	Report Notes		
Account Information C7028846 DLMP Rochest	ter	Collected 20 Aug 2013 17:00			

Iduronate-2-sulfatase, BS

Iduronate-2-sulfatase, BS

MCR

Low

<1.5 nmol/h/mL

Reference Value

≥1.5

Reviewed By

MCR

Marie Quade

Interpretation (I2SBS)

MCR

In this specimen, the activity of iduronate-2-sulfatase is reduced. This result indicates that this individual is affected with Mucopolysaccharidosis type II (Hunter syndrome; OMIM

309900), an X-linked condition. If clinically indicated, consider further confirmation by molecular genetic analysis of the IDS gene. Please note that this enzyme's activity can also be reduced in Multiple Sulfatase Deficiency (MSD; OMIM 272200). If clinically indicated, consider biochemical genetic testing of other sulfatases or molecular genetic testing of the SUMF1 gene to exclude MSD. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Fluorometric Enzyme Assay

Received: 21 Aug 2013 16:51 **Reported:** 21 Aug 2013 16:55

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905