Consider MPS VII in Perinatal NIHF

\[40\% \text{ of MPS VII patients have a history of nonimmune hydrops fetalis (NIHF)}^{1,2}\]

- NIHF is defined as abnormal fluid collection, including ascites, pleural effusions, pericardial effusions, and skin edema, in at least 2 areas of the fetus\(^3,4\).
- Lysosomal storage disorders (LSDs) were found in \(\approx 5\%\) of NIHF cases overall, and it is estimated that LSDs would be found in \(\approx 30\%\) of idiopathic NIHF cases if a comprehensive LSD workup was performed\(^4\).
- NIHF is the most common presentation of MPS VII\(^1,5\).
  - MPS VII is a rare, progressive, autosomal recessive disease characterized by \(\beta\)-glucuronidase enzyme deficiency caused by a mutation in the \(GUSB\) gene\(^1,2,6\).
- Lysosomal enzyme testing is recommended in idiopathic NIHF\(^4,7\).

Pre- and neonatal testing for MPS VII can lead to diagnosis and opportunity for early intervention\(^4\).
Additionally, if your patient tests positive for MPS VII, his or her parents will be offered access, at no cost, to a genetic counselor to address any questions or concerns by phone.