Consider MPS VII in Perinatal NIHF

≈40% 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 ≈5% of NIHF cases overall, and it is estimated that LSDs would be found in ≈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 β-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]

MPS VII = mucopolysaccharidosis type VII.

No-cost enzyme deficiency testing for MPS VII

Enzyme deficiency testing for MPS VII is available at no cost

Email **TestingforMPSVII@ultragenyx.com** for more information about testing for MPS VII

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

References: 1. Vervoort R, Islam MR, Sly WS, et al. Molecular analysis of patients with β-glucuronidase deficiency presenting as hydrops fetalis or as early mucopolysaccharidosis VII. *Am J Hum Genet*. 1996;58(3):457-471. **2. Montaño AM, Lock-Hock N, Steiner R, et al. Clinical course of sly syndrome (mucopolysaccharidosis type VII). J Med Genet. 2016;53(6):403-18. 3.** Santolaya J, Alley D, Jaffe R, Warsof SL. Antenatal classification of hydrops fetalis. *Obstet Gynecol*. 1992;79(2):256-259. **4.** Gimovsky AC, Luzi P, Berghella V. Lysosomal storage disease as an etiology of nonimmune hydrops. *Am J Obstet Gynecol*. 2015;212(3):281-290. **5.** Whybra C, Mengel E, Russo A, et al. Lysosomal storage disorder in non-immunological hydrops fetalis (NIHF) - more common than assumed? Report of four cases with transient NIHF and a review of the literature. *Orphanet J Rare Dis*. 2012;7:86. **6.** Fox JE, Volpe L, Bullaro J, Kakkis ED, Sly WS. First human treatment with investigational rhGUS enzyme replacement therapy in an advanced stage MPS VII patient. *Mol Genet Metab*. 2015;114(2):203-208. **7.** Norton ME, Chauhan SP, Dashe JS. Society for Maternal-Fetal Medicine (SMFM) clinical guideline #7: nonimmune hydrops fetalis. *Am J Obstet Gynecol*. 2015;212(2):127-139.

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