

## Child with Splenomegaly

**Q01. False positivity in beta glucosidase enzyme and genetic test came as HLH type 1, & Question 2: Beta glucosidase enzyme was positive and genetic test came as HLH type 1. How..any association:**

- No association between beta glucosidase and HLH- **the genetic report was reported as likely pathogenic/pathogenic- may take it a HLH type 1 provided the phenotype concurs well ; also possibly the false enzyme positivity with DBS may be due to a degraded sample for that matter** (even though DBS has a sensitivity of around 80 - 90%)
- Always test another enzyme as control (eg sphingomyelinase) along with glucosidase as enzymes can get degraded and give false low values

**Q02. Is Gauchers disease more common than mucopolysaccharidoses among the lysosomal disorders?**

- Gaucher disease is the commonest lysosomal storage disorder