Child with Splenomegaly

Q01. False positivity in beat 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 mucopoly sacharidoses among the lysosomal disorders?

- Gaucher disease is the commonest lysosomal storage disorder