

**Abstract ID:-** 250

**Abstract Topic:-** Clinical Genetics

**Abstract Title:-** Clinical profile and outcome of genetically confirmed glycogen storage diseases from Genetic Clinic in Marathwada, India

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**Aims:-** Glycogen storage disease are a single gene disease which are of 19 types and all have autosomal recessive inheritance except GSD type IX which can be X linked recessive. Some are liver glycogenesis like type 1, 3, 4 and 6, muscle glycogenesis like type 5, 7 and 10. some are mixed glycogenesis like type 2, 15 etc. This disease results from deficiencies of various enzymes or transport proteins in the pathway of glycogen metabolism. We present data of various types GSD diagnosed on genetic testing at Genetic clinic of MGM Hospital.

**Methods:-** Patients with massive hepatomegaly, with fasting hypoglycemia, hyperlipidemia, hyper lactatemia with CPK test were suspected to be glycogen storage disease. These patients were offered whole exome testing for confirmation of diagnosis.

**Results:-** Out of 13 cases who were on regular follow up for GSD, 7 opted for genetic testing. Three patients with GSD type VI (2 patients with PYGL gene, homozygous pathogenic c.1A>G variant and another with c.1982C>A, homozygous VUS), one patient with GSD type III (Homozygous pathogenic in AGL gene c.4260-1G>A -), 1 patient with GSD type I a (G6PC gene homozygous VUS c.182T>A) 1 with GSD type Ib (SLC37A4 c.92\_94delTCT likely pathogenic) and 1 patient with GSD type IX (PHKG2 gene, homozygous VUS c.459T>G). One patient was diagnosed as GSD based on liver biopsy but exome sequencing was negative. There were 4 males and 3 females with all of them being born consanguineously.

**Conclusions:-** The genetic confirmation of the diagnosis is given for better prognostication of disease and with better follow from the parent side, there is better understanding of the disease process and can be used for prenatal diagnostics in future pregnancies.

**Keywords:-** Abbreviation- VUS- variant of uncertain significance

Glycogen storage diseases, liver glycogenesis, muscle glycogenesis, inborn errors of metabolism.

