**Abstract ID: -** 175

**Abstract Topic: -** Clinical Genetics

**Abstract Title: -** Novel compound heterozygotes of LAMA2 variants led to demyelination of white matter in Merosin deficient muscular dystrophy (MDC1A)

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**Aims: -** In this study we aimed to understand clinical, genetic, computational analysis of two novel mutations in two children born to consanguineous couple.

**Methods: -** Genomic DNA from the peripheral blood samples was sequenced by clinical exome sequencing and confirmed by sanger sequencing. MRI and Muscle biopsy- Immunohistochemistry for demyelination and deficiency of merosin.

**Results: -** clinical exome sequencing results revealed two novel frame shift variants c.1823_1824delAT, c.5838delA in LAMA2 gene in heterozygous condition and were confirmed by Sanger's sequencing method. Immunohistochemistry and MRI which showed deficiency in merosin and demyelination of white matter. In silico analysis of this novel mutation revealed incomplete synthesis and functionally unstable, truncated protein which subsequently undergoes degradation. This is in agreement with immunohistochemistry and MRI which showed deficiency in merosin and demyelination of white matter.

**Conclusions: -** The novel mutation affected the structure and function leading to the deficiency of laminin protein.

**Keywords: -** clinical exome sequencing results revealed two novel frame shift variants c.1823_1824delAT, c.5838delA in LAMA2 gene in heterozygous condition and were confirmed by Sanger's sequencing method. Immunohistochemistry and MRI which showed deficiency in merosin and demyelination of white matter. In silico analysis of this novel mutation revealed incomplete synthesis and functionally unstable, truncated protein which subsequently undergoes degradation. This is in agreement with immunohistochemistry and MRI which showed deficiency in merosin and demyelination of white matter.