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Abstract Topic:- Clinical Genetics

Abstract Title:- Congenital Myasthenic Syndrome with Sotos syndrome

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Aims:-Congenital Myasthenic Syndromes (CMS) are a diverse group of inherited disorders caused by neuromuscular junction transmission defects. They typically present with variable weakness but intact cognition. This is a case of a girl with a dual diagnosis.

Methods:- An infant girl born prematurely at 35weeks gestation presented with floppiness and persistent oxygen dependance. She was the first child of her consanguineous parents and weighed 1.78 kg at birth. A trial of pyridostigmine helped wean her off oxygen and she was discharged. Her initial genetic panel was negative for CMS and so pyridostigmine was weaned off. Subsequently, she developed ptosis and apnea at 9 months with subtle dysmorphism. Pyridostigmine was reintroduced and she improved.

Results:- A Single Fiber Electro Myogram (SFEMG) and Repetitive nerve stimulation (RNS) performed after stopping pyridostigmine was suggestive of a neuromuscular transmission dysfunction. Over the next few years, her anthropometric parameters rapidly increased along with cognitive and learning issues. This raised the possibility of another concomitant condition. Further genetic testing was done at 4 years of age as the family was planning for a second pregnancy. Whole exome sequencing revealed two variants – one in the NSD1 gene suggestive of Sotos syndrome and another in the LAMA5 gene which was phenotyped for a presynaptic CMS. She went on to develop scoliosis which is being managed conservatively with orthosis.

Conclusions:- Genetics in CMS can be challenging at the first instance especially when genotypic phenotypic correlation is difficult. Consider the possibility of a dual diagnosis when the clinical picture does not fit.

Keywords:- Myasthenic Syndrome, Sotos syndrome