DOK-7 DECOY : WHERE MYASTHENIA HIDES IN PLAIN SIGHT

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BACKGROUND & AIMS :

- ✓ DOK-7 is a postsynaptic cytoplasmic protein that binds and activates muscle specific tyrosine kinase, which inturn leads to Rapsyn-associated endplate AchR clustering and normal folding of the post synaptic membrane.
- ✓ The Typical phenotype of DOK-7 myasthenia is of a limb girdle weakness with non specific myopathic features. Useful differentiating features include : Onset after infancy, normal walking age milestone, limb girdle phenotype, normal eye movements, stridor, tongue wasting, associated non-specific myopathy and poor response to pyridostigmine and 3,4 – di-aminopyridine.

METHODOLOGY :

Herewith we present a case report of patient presenting with DOK 7 congenital myasthenia

CASE REPORT :

- 35 year old female patient, with normal developmental milestones, presented with insidious onset difficulty in using both proximal upper limbs for the past 13 years, difficulty in getting up from squatting position for the past 13 years. drooping of bilateral eyelids, for the past 12 years, difficulty in breathing and worsening of bilateral upper and lower limb weakness for 3 months
- On examination, bilateral ptosis was present and restriction of extraocular movements in all directions, no tongue wasting or atrophy, bilateral palatal movements equal. Tone was normal in all 4 limbs. Proximal power was 3/5, distal power was 4/5 in all 4 limbs. Patient was tachypnoeic, requiring CPAP support. Routine investigations were normal and serum CPK was normal.
- Whole exome sequencing with mitochondrial gene sequencing was sent and patient was found to have to have DOK 7 mutation positive. Patient was started on oral salbutamol, following which she improved

RESULTS : Here we have discussed a case report of a 35 year old female, presenting with bilateral ptosis and limb girdle onset weakness, progressing to distal muscles, along with respiratory involvement, considering the above presentation, differential diagnosis of congenital myopathy, congenital myasthenia, mitochondrial myopathy was suspected

- Features favouring DOK-7 congenital myasthenic syndrome include : Onset after infancy, normal walking milestones, limb girdle phenotype and good response to salbuatmol. Features contradicting DOK-7 congenital myasthenic syndrome include complete ophthalmoparesis and absence of tongue wasting.

DISCUSSION AND CONCLUSION : DOK7 congenital myasthenic syndrome tends to present at a later age and in DOK7 CMS eye movements are usually spared and anticholinesterases can exacerbate the weakness. The presence of stridor in early onset cases and the observation of tongue wasting may be specific clues. Treatment with ephedrine or oral salbutamol can result in a slow, steady, and often dramatic improvement over months. Hence a high index of clinical suspicion is necessary for diagnosis and appropriate management of the disorder.