A rare case of dystonia in siblings due to SCO2 mutation.

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ABSTRACT - A 12-year-old girl presented to the neurology department with complaints of difficulty in walking for 3 years. Parents noticed slurred speech and posturing of fingers and toes while walking since 2years. She was born of consanguineous parentage. She had normal motor and mental milestones. The elder sibling male 14 years developed similar complaints from 5 yrs of age. Now he is severely dysarthric and unable to walk. On examination the girl was dysarthric with severe dystonia of both upper and lower limbs and needed support for walking. Laboratory investigations revealed elevated lactate with normal ceruloplasmin level. MRI brain was normal. She was treated with L-Dopa with no improvement. In view of consanguinity and prolonged history, genetic etiology was considered. Plasma was sent for whole exome sequencing, and it revealed SCO 2 gene mutation. This gene mutation causes dysfunction of mitochondrial respiratory chain complex IV thus causing dystonia and other extrapyramidal symptoms

This is first reported case of dystonia due to SCO2 mutation from India.

SCO2 is a mitochondrial copper binding protein important for mitochondrial respiratory chain type IV complexes. In addition, SCO1 and SCO 2 may also play a role in cellular homeostasis as mutations have been shown to result in severe copper deficiency in affected tissues. As the clinical picture in this case is similar to Wilson's disease, copper deficiency in cerebral tissues may be one of the reasons for dystonia. Further studies are needed to know the relation between SCO2 mutation and copper homeostasis in the brain. We also be be aware of SCO2 mutations as one of causes of severe extrapyramidal symptoms in children for further genetic counselling.

Biography.

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MBBS from Guntur medical college Guntur India (1991-1996)

MD (internal Medicine) from Guntur medical college (1997-2001)

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