INTRODUCTION:

Vitamins used in clinical practice as food supplements sometimes act as cofactors for catalysis of many metabolic processes in the body. A few neurological disorders, including seizures, have been proved to be treated with vitamins in children. Progressive bulbar palsy in infants is a severe debilitating illness with high morbidity. Many of the etiologies of the disorder seem to be mostly neurodegenerative and have poor outcomes. Fazio-Londe disease, a rare disorder with progressive bulbar paralysis and respiratory failure, is found to be due to mutations in the SLC52A3 gene which encodes the intestinal (hRFT2) riboflavin transporter in children. They respond very well to prompt riboflavin therapy as in the case we described and thus emphasizing this entity of riboflavin responsive neurological disorder as a unique vitamin responsive problem.

CASE HISTORY:

A 7-months-old female infant presented with swallowing difficulty, poor cry and pooling of secretions of the throat as progressive bulbar palsy. She responded to riboflavin treatment and continued to improve.

Key-words: bulbar palsy, riboflavin, hypotonia, respiratory distress

Key Messages: riboflavin responsive bulbar palsy.
investigations. MR brain and cervical spine were normal and metabolic screening with tandem mass spectroscopy, vitamin B 12 levels, survival motor neuron gene PCR (SMN gene) for spinal muscular atrophy were all negative. Detailed nerve conduction study revealed normal conduction velocities and F waves, normal sensory potentials; needle electromyography (EMG) showed fibrillation potentials over deltoid, biceps and triceps on both sides revealing axonal neuropathy (anterior horn cell). Parents refused for muscle biopsy and CSF study to this child. With the limited available clinical picture and electro-physiological investigations she was diagnosed as neurogenic bulbo-spinal amyotrophy probably due to anterior horn cell disease. Following possibilities were put forth: spinal muscular atrophy (SMN gene mutation negative), hereditary motor sensory neuropathy type 1 (nerve conduction study normal), Brown-Vialetto-Van Laere syndrome (normal hearing), Tay Sach's disease (absent startle myoclonus, cherry red spot) Fazio Londe disease. With the above differential diagnosis, she child could be suffering from progressive bulbar palsy of Fazio Londe disease; a trial of riboflavin treatment at a dose of 15 mg per Kg body weight was given. Surprisingly, she showed dramatic improvement in 2 weeks time and successful extubation was done on the 25th day. Riboflavin was then continued on 10 mg per Kg weight of the child and discharged from hospital with the same medication. She was reviewed after 3 months; she started to swallow liquids as well semi solid food with no nasal regurgitation, could hold the neck and could sit and stand with support. She is still on the above treatment and advised regular follow up.

**DISCUSSION:**

Fazio Londe disease is a rare neurological disorder presenting with progressive bulbar palsy with respiratory failure. (1) It was once considered to have an unrelenting course, is now found to be due to mutations in the SLC52A3 gene which encodes the intestinal (hRFT2) riboflavin transporter in some children (2). Brown-Vialetto-Van Laere syndrome is similar rare neurological disorder with sensory-neural deafness, presenting at all ages with bulbar palsy and respiratory compromise and Fazio-Londe syndrome is considered to be the same disease entity presenting in children with normal hearing. (3) Appropriate diagnosis requires mutation analysis of all the related three transporter genes of the above. (4) Because of the striking and often lifesaving effects of riboflavin supplementation it is highly advisable to start treatment immediately without awaiting results of mutation analysis. Thus despite lack of availability of genetic analysis, a trial of riboflavin therapy, administered to our child, on the basis of clinical symptoms resulted in a life saving effect.

**CONCLUSION:**

This case illustrates the rarity of presentation of progressive bulbar palsy of Fazio Lande disease in this child on clinical and limited electrophysiological investigation and the beneficial life saving role of riboflavin treatment for this disorder. The point of philosophy is to start with something so simple and to end with something so surprising that no one will believe it. Points to ponder!

**REFERENCES:**


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