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A Rare Case of Progressive Bulbar Palsy of Childhood

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Abstract: ***Progressive bulbar palsy of childhood are a spectrum of disorders which encompasses diseases like Fazio londe syndrome, Brown vialetto Van laere syndrome (BVVL), Nathalie syndrome, boltshauser syndrome and others. Fazio londe syndrome is commonest characterized by progressive bulbar palsy with weakness of limbs[1], BVVL has similar features associated with sensorineural deafness. Nathalie syndrome has additional features like cataract, cardiac conduction defects and hypogonadism. Boltshauser syndrome has SNHL and isolated vocal cord paralysis. However, none of these syndromes have facial dysmorphism. Here we present a case of childhood anterior horn cell disease with facial dysmorphism[2].***

Keywords: Fazio Londe syndrome, progressive bulbar palsy, facial dysmorphism, childhood motor neuron disease, sensorineural hearing loss

1. Case Report

A 13 year old male child has swallowing difficulty and weakness of the limbs since last five years. His mother noticed that the child has difficulty in chewing food and swallowing both solids and liquids with frequent oral and nasal regurgitations of the swallowed food particle at nights. Mother also has complained of low volume hypernasal speech which is improving on bending forward and difficulty in spitting water, difficulty in sipping through the straw, difficulty in holding objects and lifting them overhead and difficulty in getting up from the squatting position and difficult in gripping onto the slippers tightly. No myalgias, myotonia, cold aggravating weakness, muscle twitching, diurnal variation of symptoms. He is born out of third degree consanguineous parentage, second in birth order, normal delivery with no birth asphyxia, normal developmental milestones. There is no history of similar complaints in the family. On general examination the child is alert with normal vitals and no organomegaly with facial dysmorphism with hypertelorism, low set ears, high arched palate, elongated face, genu valgum. Nervous system examination showed normal intellect with MMSE 30/30. Cranial nerve examination showed normal vision, normal visual fields, color vision and normal fundus, no optic atrophy/retinitis pigmentosa, muscles of mastication and facial muscles are weak, decreased palatal movements decreased tongue movement with atrophy and fasciculations (video 1). Motor system examination showed generalized wasting, normal tone, power of 4/5 in all four limbs with hyperreflexia. There is waddling gait. Rest of the nervous system examination is normal. Investigations revealed normal CBP, RFT, LFT, Thyroid profile, Vitamin B12 levels, serum cortisol, serum ACTH. Nerve conduction studies showed sensory motor axonal neuropathy, MRI brain with cervical spine was normal, Schirmer test done indicative of lacrimatory failure. Pure tone audiometry was normal. ACTH stimulation test is normal. Barium swallow showed bird beak appearance (figure 1) suggestive of achalasia Upper GI endoscopy and manometry studies showed presence of Achalasia with no visual peristaltic moments (Figure 3). Genetic analysis for ribosomal transporter protein genes and ALADIN gene was sent which came to be negative. With the help of Gastroenterologist patient was kept on peg tube and esophageal dilatation procedure was done and patient is kept on follow up.

2. Discussion

Allgrove syndrome is rare disorder with prevalence of one in one lakh individuals[3]. It is characterized by presence of achalasia, adrenal crisis, alacrimia and amyotrophy. Adrenal crisis will occur in most of the patients but in our case it is not present or not yet manifested. Achalasia can be managed with laparoscopic hellers myotomy, pneumatic esophageal dilatation, etc. Alacrimia is managed with artificial tears to avoid complication like keratitis. Recurrent respiratory infections are common and has to be treated with antibiotics.

3. Conclusion

Our case has incomplete presentation of Allgroove syndrome without adrenal insufficiency and presence of bulbar weakness and achalasia. This case demonstrates difficulty in diagnosis as characteristic triad is not always seen and may delay the diagnosis.

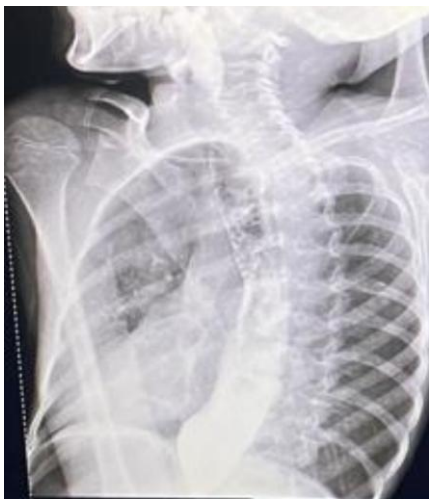

Figure 1: Barium swallow study showed bird beak appearance suggestive of achalasia

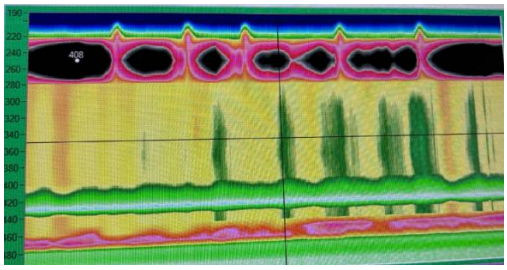

Figure 2: Esophageal manometry studies showed no visible peristaltic movements

tongue fasciculations.mp4

Video 1: Demonstrating tongue fasciculations and atrophy

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