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Case Report

Rare Case Primary Report: Xia Gibb's Syndrome

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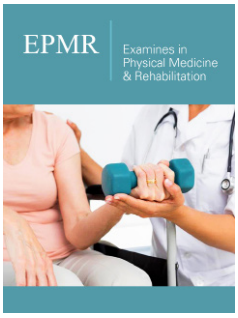

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Abstract

Xia-Gibbs syndrome (Mental retardation, autosomal dominant 25; MRD25) (MIM 615829), first described in 2014, is a rare autosomal dominant disease characterized by mental retardation, developmental delay, speech delay, structural brain anomalies, hypotonicity, protuberant eyes, visual problems, laryngomalacia and low sleep quality. It occurs due to heterozygous mutations in the AT-hook DNA binding motif containing 1 (AHDC1) gene on chromosome 1p36. Research reports published in 2020 by Ping et al. [1], states that about 100 patients worldwide have been identified, all of them in the pediatric age or adulthood. To date, no fetal XGS has been reported [1-4]. Xia-Gibbs syndrome (XGS) is a recently described neurodevelopmental disorder due to heterozygous loss-of-function AHDC1 mutations. XGS is characterized by global developmental delay, intellectual disability, hypotonia, and sleep abnormalities. We are here to report our primary observations and findings of a diagnosed case of Xia Gibb's Syndrome referred for Physiotherapy with signs of development delay, atonia, joint laxity and related features.

Keywords: Xia-Gibbs syndrome; XGS; Physiotherapy

Introduction

Eleven months old Ayesha Sajeer, a diagnosed case of "Xia Gibb's Syndrome", (XGS) was referred for Physiotherapy in the second week of April 2021 through the Physical Medicine and Rehabilitation Department of Government District Hospital, Tirur, Kerala. She was sent with signs of global development delay, dysmorphism and poor muscle tone. Baby Ayesha was born as the 3rd child for a non-consanguineous couple without any significant prenatal and perinatal events. She was born as NVD, cried immediately and the recorded birth weight was 2.650Kgs. No indications for immediate intensive care were noted on day one. However, on the second day evaluation, the Pediatrician noticed a typical cardiac murmur which was later confirmed as CHD - Atrial Septal Defect. This was managed conservatively from then.

Baby Ayesha was presented with general motor development delay, hypotonia, peripheral joints laxity, high forehead, hypertelorism, bilateral hypo-pigmented iris, depressed nasal bridge, upturned nares, low set ears, smooth philtrum, thin and tented upper lip, short neck, transverse palmar crease, bilateral clinodactyly, oedematous limbs (specially feet), incurving 5th toes (bilateral) and mild hearing loss bilaterally. No symptoms of sleep apnea, tracheomalacia, seizures, strabismus and orthopedic deformities were noted. Patient is too young to evaluate autistic signs, intellectual disabilities, speech delays, sensory processing disorders and behavioral issues. Brain MRI was not available to assess any structural brain defects. Mother is 29 years old now, homemaker and father are 39 years, working abroad as an unskilled labourer. Siblings are growing normal and are aged 9 and 4 years. No cases like this were reported both on the maternal and paternal side. Baby Ayesha was the first case of Xia Gibb's Syndrome (XGS) attended by our department for Physiotherapy services. This is the first case reported in this region. After the basic clinical evaluation on the day, the case was asked to come on a later date to start with therapeutic interventions.

Meanwhile, we did refer to the available details on the web. Since this was a newly identified genetic disorder, not many articles were found on this. Only one article was found during our search done between 13/04/2021 & 21/04/2021. This was published from India by Goyal C, Naqvi W, Sahu A in 2020, at Cureus 12(8). According to Goyal et al. [5] "NDT and SI approaches along with the use of appropriate orthoses accelerated the achievement of motor milestones in their case." However, the case seen by them was a 27 month boy with difficulty in sitting without support. Based on our clinical experiences in Pediatric Physiotherapy and the little information we have gathered from the literature, we have taken this case for institution-based habilitation. We have enlisted the following where Physiotherapy interventions are indicated. (1) Global developmental delay (2) Hypotonia and joints laxity (3) Spine care as precautionary (4) Orthotic supports as SOS. We believe a regular therapist guided NDT program comprising weight shifts, transitions, posturing and movement facilitation along with accommodative chair, vestibular & reticular tracts stimulations, weight bearing acts etc will help her progress.

Presently, we have scheduled 4/7 Physiotherapy visits for her. Gradually, once I gain the confidence of the parents and vice versa, the exercise regime can be progressed to home based with periodic reviews. Speech therapy, Audiology and Occupational therapy consultations will be scheduled as and when required. A therapy diary to record the day-to-day events, medical and therapy follow etc. and also comprising all exercises to be followed at home, is issued to her. The status of her progress, outcome measures, procedural images etc (Figure 1 & 2), will be reported after 8-12 months of interventions.

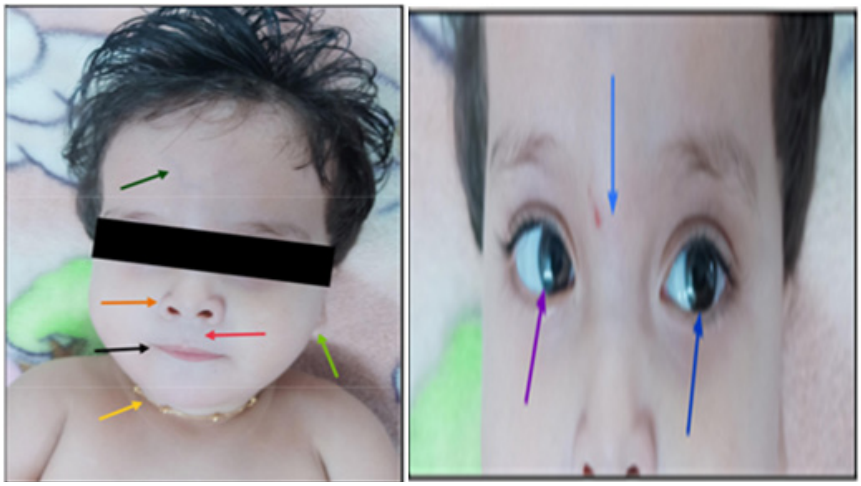

Figure 1: Features of dysmorphism.

Figure 2: Transverse palmar crease.

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