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Charge syndrome: A case report

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Abstract

Charge syndrome is an uncommon condition defined by a number of congenital abnormalities that are inherited in an autosomal dominant way. We provide a case report to improve physician experience and understanding of Charge syndrome diagnosis using clinical diagnostic criteria. A 10-year-old male child was admitted with complaints of bluish discoloration of fingers and lips beginning at 1 year of age, weakness of the left side of the body beginning at 4 months, and involuntary movements of the left upper limb beginning at 2 months. In our instance, the patient received symptomatic medical care, surgical care for heart abnormalities and dysmorphic characteristics was provided with parental counselling, and physical and occupational therapy was provided. According to the current case report, the diagnosis of isolated dysmorphism necessitates a comprehensive examination and research to check for syndromic connection.

Keywords: Charge Syndrome, Coloboma, Cyanosis, Clubbing, CHD7.

Introduction

Charge syndrome is an uncommon condition defined by a cluster of congenital abnormalities that is inherited autosomal dominantly [1]. The prevalence of Charge syndrome ranges from 1:8,500 to 1:15,000, with the majority of cases being sporadic [2, 3]. Given the present global incidence, it is fair to assume that there is a significant percentage of underdiagnosis due to poor clinical recognition during the newborn era. Because of the wide variety of systems impacted, managing Charge syndrome is difficult. Typically, many physicians are

engaged, and children attend repeated, frequently fragmented outpatient appointments. As a result, we provide a case report to improve physician experience and understanding of Charge syndrome diagnosis using clinical diagnostic criteria.

Case Presentation

A 10 years old male child born of non-consanguineous marriage admitted with complaints of bluish discolouration of fingers and lips noticed from 1 year of age, weakness of left side of body from 4 months and involuntary movements on left upper limb from 2 months. Gradual increase in bluish discoloration from past 6 months associated with decrease in playfulness and normal routine activity of the child. Child had history of multiple episodes of sudden increase in bluish discoloration of body along with restlessness which was relieved on sitting in palthi position. 4 years back child was operated for right sided inguinal hernia.

On examination, the patient was conscious and oriented with Pulse rate - 108 bpm, Bp-112/70 mm of Hg in RAS. No significant difference was noted in all 4 limbs. Child had central & peripheral cyanosis (SpO2-79% in room air) along with grade 3 Clubbing (Figure 1). The body weight was 16.1kg (<3SD), the height was 116 cm (< 3SD). Head-to-toe examination showed bilateral Iris coloboma at 6'clock position, Microphthalmia with microcornea, bilateral epicanthal fold and hypertelorism, depressed nasal bridge, low set ears with external ear abnormality and deviation of angle of mouth to right, High arched palate, dental malocclusion, Micropenis (SPL-3cm) and bilateral undescended testis. Choreo-Athetoid movements were seen.

Figure 1: Physical Examination of patient

A: Antihelix discontiniuous with the antitragus, prominent inferior antihelix, notch between Antihelix and antitragus. B: Iris Coloboma. C: Clubbing. D: Facial examination

Nervous system examination revealed that the patient had decreased word output with incomprehensive speech and left UMN Facial nerve palsy. On the basis of above mentioned examination findings, an impression of Charge syndrome was made.

2D Echo showed tetrology of fallot(TOF). MRI Brain was normal. USG abdomen showed bilaterally undescended testis (right sided noted just below superficial inguinal ring and left sided in inguinal canal). Chest X-ray confirmed that pulmonary oligemia and boot shaped heart (Figure 2). Patient was managed with partial exchange transfusion and referred to cardio thoracic surgeon for planning of cardiac surgeries.

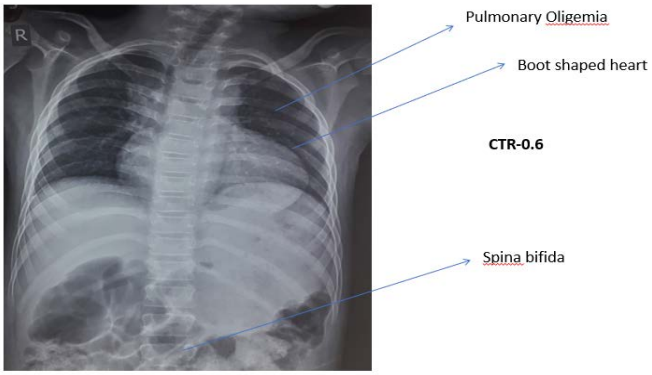

Figure 2: Chest X-ray of patient

Discussion

Charge syndrome is mostly caused by a heterozygous mutation in the CHD7 gene on chromosome 8q12, which was identified in 2004 by Vissers et al [4]. Over 90% of Charge patients who fulfil Blake et al and/or Verloes' clinical diagnostic criteria have the CHD7 mutation [5]. When all of the test findings were added together, our patient was diagnosed with typical Charge syndrome. However, the Charge syndrome is characterized by a wide range of symptom and finding combinations. Blake and Prasad [6] reported the incidence of phenotypic features which includes Developmental Delay~100%, Ear anomaly-80-100%, Coloboma-80-90%, Cardiovascular Malformation-75-85%, Growth retardation-70-80%, Coanal Atresia-50-60%, Genital hypoplasia-50-60% and Cranial nerve -40-90%. In our case, Colaboma, Ear Anomaly, Cranial Nerve palsy (7th), cardiovascular anomaly (TOF), Developmental Delay, Growth retardation, and Genital hypoplasia (Micropenis, Undescended Testis) was present. Blake criteria and Varloes criteria of our patient are depicted in table 1.

Parameter	Blake Criteria	Varloes Criteria
Major	3	1 (2)
Minor	4	5
Interpretation	Definite	Atypical (Typical)

The most frequent newborn emergencies with Charge syndrome are cyanosis owing to congenital cardiac abnormalities or bilateral posterior choanal atresia, with trachea-esophageal fistula being less probable [7]. As a result, all individuals suspected of having Charge syndrome should consult a cardiologist. Prostaglandin should be administered to the child if he or she has patent ductus arteriosus and restricted pulmonary blood flow. Tracheostomy is required in some situations to treat persistent airway problems, aspiration, or gastroesophageal reflux illness. Children with Charge syndrome require significant medical treatment for their eating problems, which necessitates the use of jejunostomy or gastrostomy feeding tubes on a daily basis [8]. Intubation may be problematic in children with Charge syndrome. As a result, for scheduled surgical operations, a pediatric otolaryngologist or anesthesiologist should be present. Artificial tears can help individuals with facial palsy avoid corneal scarring. Hearing aids should be worn as soon as hearing loss is identified, with frequent monitoring [9]. In our case, the patient received symptomatic medical care, surgical care for heart abnormalities and dysmorphic characteristics was provided with parental counseling, and physical and occupational therapy was provided. According to the current case report, the diagnosis of isolated dysmorphism necessitates a comprehensive examination and investigations to check for syndromic connection.

Conclusion

An inter professional team that provides a comprehensive and cohesive approach to service delivery can provide the greatest outcomes for a patient with Charge syndrome. Early diagnosis of the condition and diagnostic workup to identify specific symptoms will aid in the prevention of related problems. Charge syndrome is a complex condition that necessitates consultation with a geneticist, pediatric cardiologist, pediatric otolaryngologist, or anesthesiologist in order to reach a consensus decision that will result in better outcomes.

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