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CASE REPORTS

AN UNUSUAL CASE OF CHARGE SYNDROME PRESENTING WITH INTRATHORACIC KIDNEY AND RIGHT-SIDED DIAPHRAGMATIC HERNIA

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ABSTRACT

CHARGE syndrome is a complex genetic disorder due to mutations in the CHD7 gene. Congenital diaphragmatic hernia has been rarely described in CHARGE syndrome. The following report describes an atypical presentation of this rare syndrome in a male infant associated with a right-sided Bochdaleck-type diaphragmatic hernia and intrathoracic right kidney.

ARTICLE HISTORY

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KEYWORDS

diaphragmatic hernia, intrathoracic kidney, CHARGE syndrome, neonate

Introduction

CHARGE syndrome is a rare heterogenous syndrome with multiple congenital anomalies and a prevalence of one in 10 000. (1) The acronym CHARGE is based on the cardinal features identified when the syndrome was delineated: Coloboma, Heart malformation, Choanal Atresia, Retardation of growth and/or development, Genital anomalies, and Ear anomalies. Since that time, new frequent clinical findings have been added to the clinical spectrum of this syndrome. We report a patient affected by CHARGE syndrome presenting with congenital diaphragmatic hernia (CDH) and intrathoracic trans-diaphragmatic kidney.

Case Report

A male neonate was born to non-consanguineous parents by vaginal delivery at 39 weeks of gestation. Apgar scores were 9 and 10 at 1 and 5 minutes after birth respectively. He was referred to our department immediately after birth for treatment of mild respiratory distress. Physical examination on admission showed the following: Birth weight, 2700 g (5th to 10th percentile); crown to rump length, 49 cm (25th to 50th percentile); head circumference, 34 cm (10th to 25th percentile); body temperature, 36.4°C; respiratory rate, 58 breaths per minute; pulse rate, 144 beats per minute and regular; blood pressure, 75/35 mmHg; and percutaneous arterial oxygen saturation, 94%. Sensorium, body posture and muscle tone were normal. Dysmorphic features were noted including right choanal atresia, grade III microtia (Figure 1), and square-shaped face, downward slanting palpebral fissures, micrognathia and right malar flattening leading to facial asymmetry without facial palsy. Furthermore, neither limb or thoracic deformity nor heart murmur were detected. The abdomen was soft, and no anomaly was observed in the genitalia. Ophthalmic examination was normal. Chest radiograph showed the presence of intestinal loops in thorax suggesting a right-sided diaphragmatic hernia (Figure 2). Ultrasound abdomen showed normal diaphragmatic mobility and an empty renal fossa on the right side. Thoracic computed tomography (CT) scan confirmed the presence of right-sided diaphragmatic Bochdaleck-type hernia associated with trans-diaphragmatic intrathoracic ectopic right kidney (Figure 3). Temporal bone CT on the right side showed agenesis of the external auditory canal, small opacified middle ear cleft, incudo-malleolar ankylosis, agenesis of the oval window, non-pneumatized mastoid, narrow internal auditory canal with normal appearing semicircular canals, vestibule and vestibular aqueduct. Tympanic portion of the seventh cranial nerve was not visualized. CT scan imaging of the left ear was normal. Cerebral CT confirmed the presence of membranous choanal atresia. Echocardiogram revealed patent foramen ovale and ductus arteriosus. On the basis of the above-mentioned findings the patient was diagnosed to have atypical CHARGE syndrome. Genetic testing could not be performed due to unaffordability. The respiratory symptoms resolved by 12 hours and oxygen administration was discontinued. The infant was discharged at one week of age and followed-up on an outpatient basis.

Figure 1. Grade III microtia: hypoplasia of the pinna with a small peanut-like vestige associated with atresia of the external auditory canal

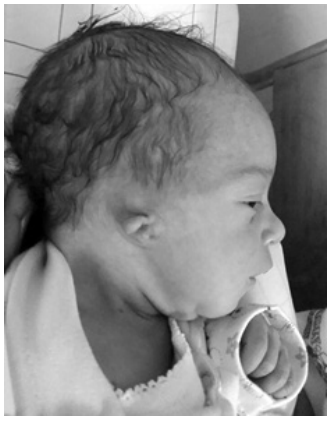

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Figure 2. Chest radiograph showing the presence of ill-defined paracadiac opacity on the right side with intestinal loops in thorax suggesting a right-sided diaphragmatic hernia

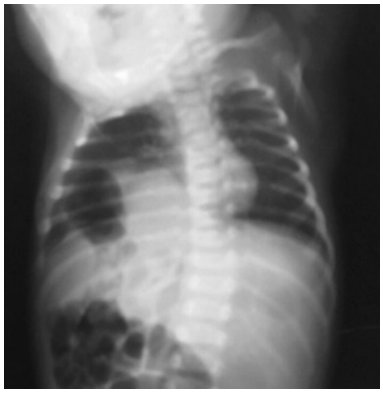
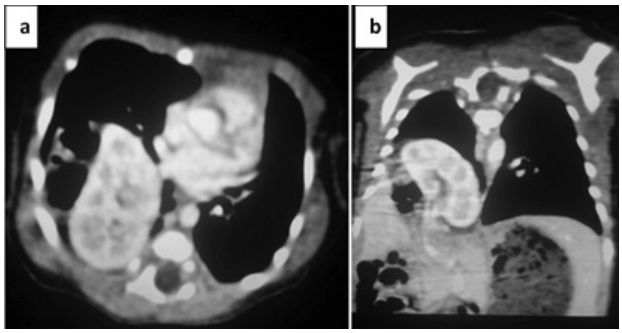


Figure 3. CT chest: (a) Axial view showing a right diaphragmatic hernia with intrathoracic bowels and right kidney (b) coronal reconstruction showing the diaphragmatic defect and herniation of right kidney and bowels into the thoracic cavity.



Discussion

The phenotypic heterogeneity of CHARGE syndrome is considerable and diagnostic criteria have been refined several times. Verloes proposed renewed clinical criteria in 2005, and gave a formal definition for partial and atypical CHARGE syndrome, the latter includes two majors but no minors or one major and two minors. (2) Our patient fulfilled diagnostic criteria for atypical CHARGE syndrome as he had choanal atresia, microtia, and patent ductus arteriosus. Furthermore, our patient presented with a right-sided diaphragmatic hernia and intrathoracic right kidney, highlighting the vast clinical variability of occasionally associated findings. (3) Only one case has been reported with such an association with CDH and none with intrathoracic kidney. (4-5) Intrathoracic ectopic kidney accounts for 5% of all renal ectopias, its diagnosis in the neonatal period is extremely rare and its association with CDH is even rarer with an incidence of only 0.25%, this condition is uncommon on the right side (10–20%). (6)

Casaccia et al. suggested on the basis of documented reports and embryogenesis studies that a disturbance of mesenchymal–epithelial interactions, where mesenchymal stands for mesenchyme and mesoderm and epithelial includes ectoderm and endoderm, could be considered the common pathogenetic mechanism in selected syndromic cases affected by CDH and CHARGE, explaining in part their co-occurrence. (5) Further studies are needed to better understand the pathogenesis of such association. Interestingly, the long arm of chromosome 8 is involved in both pathologies, suggesting the presence of important embryonic developmental genes. (5) A range of heterozygous mutations in the CHD7 gene on chromosome 8 has been detected in 60%-70%of patients with CHARGE syndrome [9], this testing has not been performed in our study given its high cost. CHARGE syndrome is, however, a clinical diagnosis. The clinical scores have shown their robustness, as almost all patients with CHD7 mutation fit them. (1)

Conclusion

Charge syndrome is not exceptional; it should be kept in mind that the mild end of the phenotypic spectrum is still expanding. Despite the extremely rare association of CDH and CHARGE syndrome, patients with CDH should be actively screened for CHARGE syndrome findings.

Compliance with Ethical Standards

Ethics statement: Informed consent was taken from the parents.

Funding: None
Conflict of Interest: None

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