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Pulmonary artery sling – A novel cardiovascular finding in a patient with distal 18q deletion

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ABSTRACT

Among the rare chromosome abnormalities compatible with life, partial monosomy of chromosome 18 is a relatively frequent chromosomal aberration. The affected individuals are at relatively increased risk of having congenital cardiac defects with the pulmonary valve anomalies and atrial septal defects being the most common findings. We report a patient with a distal 18q deletion, follow-up from birth to 15 years of age, and a novel finding of distal 18q—associated cardiovascular anomaly - a pulmonary artery sling. Since birth, the patient presented with characteristic phenotype features, respiratory distress, and frequent respiratory infections. Echocardiographic examination revealed atrial septal defect and a suspicion for presence of pulmonary artery sling, which was later confirmed using computed tomography angiocardiography. During the surgery at 18 months of age, patent ductus arteriosus was found and repaired together with repair of atrial septal defect and pulmonary artery sling. Postoperatively, the stridor and respiratory distress gradually disappeared. Since exact echocardiographic imaging of vascular anomalies can be challenging in some patients, the occurrence of pulmonary artery sling in the individuals with distal 18q deletion is likely not so exceptional, but rather it may be missed during routine echocardiographic evaluation. Therefore, the definitive diagnosis should be made using computed tomography angiocardiography, which is preferred to magnetic resonance imaging due to better visualization of lung parenchyma and airways, higher spatial resolution, and faster scanning with lower requirement of sedation. To our knowledge, this is the first report of pulmonary artery sling in a patient with a distal 18q deletion.

1. Introduction

Partial monosomy of chromosome 18 is, among the rare chromosome abnormalities compatible with life, a relatively frequent segmental aneusomy with the incidence of 18q deletion about 1:40000 livebirths [1,2]. The manifestations of this condition vary greatly between individuals and the most common clinical findings are developmental delay, short stature, seizures, abnormal behaviour, minor facial dismorphology, weak muscle tone, narrow auditory canals leading to hearing loss, various forms of intellectual disability, and cardiovascular anomalies [2–4].

We present a case of a patient diagnosed with a distal 18q deletion with the characteristic phenotype features observed from birth up to 15 years of age and a novel finding of associated cardiovascular anomaly - pulmonary artery sling. To our knowledge, this is the first report of pulmonary artery sling in a patient with distal 18q-.

2. Case Report

2.1. Clinical Status after Birth

The boy was born at the 41st week of gestation from the second

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gravidity by caesarean section (birth weight: 2140 g, height: 48 cm). After delivery, the patient presented with following clinical findings: facial dysmorphias, short stature, short neck and a round face, microcephaly, upslanting palpebral fissures, high and intact palate. Nasal bridge was flat and broad. He had drooping eyelids (ptosis), hypertelorism, and an extra fold of skin covering the inner corners of the eyes (epicanthic folds). The ears were low-set with large and prominent ear lobules. The philtrum was short and the mouth wide with down-turned corners with prominent lower lip and tented thin upper lip. Limb movements were asymmetric, syndactyly between the second and the third finger of the right foot was present, and the second finger was partly folded over the third finger. The palmar skin was thick with paucity of creases.

2.2. Applied Interventions

Cardiorespiratory distress requiring ventilatory support started soon after birth. Tracheostomy and gastric tube placement has been done in order to provide long term ventilatory support and nutrition. Echocardiographic examination revealed atrial septal defect and a suspicion for presence of pulmonary artery sling, which was later confirmed using computed tomography angiocardiography (Figs. 1 and 2). Generalized seizures were controlled with valproate.

2.3. Genetic Counselling

The first genetic consultation was done at the age of 4 months due to congenital heart disease and facial dysmorphic features. The analysis of G-banded chromosomes derived from cultured peripheral blood lymphocytes confirmed the abnormal karyotype 46,XY,del(18)(q21). Since the karyotypes of both parents were normal, the 18q deletion arose de novo.

2.4. Follow-up

Since birth, the patient suffered from respiratory distress, frequent respiratory infections, and pneumonias. Atrial septal defect and pulmonary artery sling were repaired at 18 months of age. Patent ductus arteriosus was found during the surgery and was closed at that time. Postoperatively, the stridor and respiratory distress gradually disappeared. The patient is currently a 15-year-old boy with asthenic habitus, characterised by decreased anthropometric parameters below the third percentile of reference range – weight 26 kg, height 126 cm. The patient suffers from deformed chest and kyphoscoliosis, and he has been diagnosed with hypothyroidism. Global delay in brain maturation results in impaired cognitive, motoric and emotional abilities including speech problems, cognitive dysfunction affecting attention, concentration and others. Gross and fine motor milestones are severely delayed. Furthermore, intellectual functioning is at the level of severe intellectual disability without complex communication skills. Nutrition is supplied through a gastrostomy tube. Toilet training is not present. Oxygen is supplied via tracheostomy intermittently during day and at night. Personal assistant services for all activities of daily living are needed.

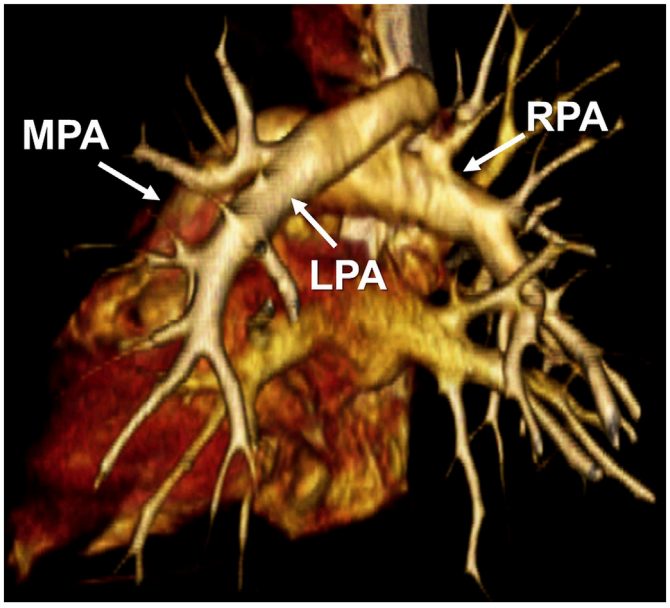

Fig. 1. Three-dimensional virtually reconstructed computed tomography image of the left pulmonary artery sling - the lateral and posterior view. MPA – the main pulmonary artery, RPA - the right pulmonary artery, LPA – the left pulmonary artery. Philips Brilliance CT, 40 slice, 140 kV.

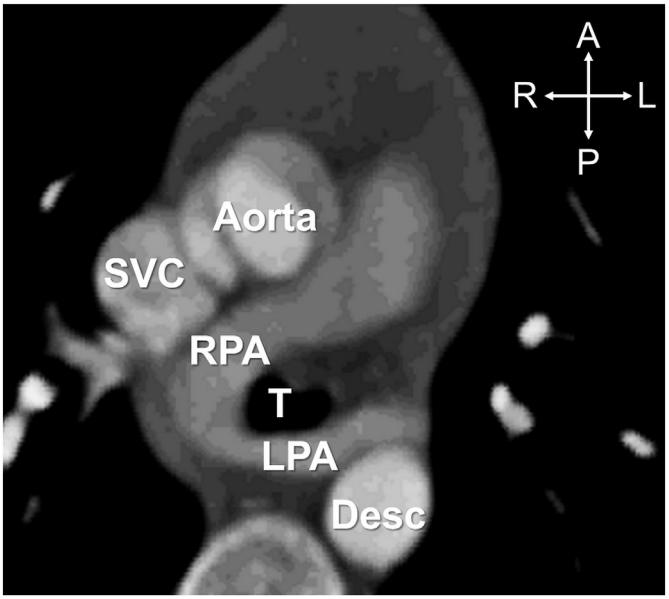

Fig. 2. Computed tomography axial image. The left pulmonary artery (LPA) arises in anomalous fashion from the right pulmonary artery (RPA) and is located behind the trachea (T) causing its compression. SVC – superior vena cava, Desc – descending aorta. Philips Brilliance CT, 40 slice, 140 kV.

3. Discussion

Distal 18q deletion is a well-described disorder resulting from a partial deletion of the long arm of chromosome 18 [5]. Suspicion of the chromosomal abnormalities is higher in the presence of birth defects or developmental delay. To effectively diagnose this condition, a genetic analysis needs to be performed. However, the size and regions of the deletion are highly variable, what may lead to large differences in the severity and high variability of the symptoms of distal 18q- [4].

Although congenital cardiac anomalies are not the common characteristic consequences of distal 18q deletion, the affected individuals are at relatively increased risk of having cardiac defect with the incidence approximately 29% (ranging from 24% up to 54% in distinct studies) [2,4–6]. No single specific cardiac defect seems to be dominant, but the pulmonary valve anomalies and atrial septal defects were the most commonly detected findings [6]. Therefore, a detailed cardiovascular examination consisting of physical examination, electrocardiography (ECG) and ultrasound examination is highly recommended in this group of patients [6].

The patient presented in this study has a severe form of the distal 18q- with almost all characteristic features of this condition, as well as cardiac abnormalities: atrial septal defect, patent ductus arteriosus, and

pulmonary artery sling, which was reported, to our knowledge, for the first time in association with distal 18q deletion. Pulmonary artery sling is a rare congenital vascular anomaly that typically presents during infancy with severe respiratory symptoms, stridor, and respiratory distress, which could be exacerbated by infections, similarly as in our patient [7]. After closure of the atrial septal defect and ductus arteriosus, and reconstruction of the left pulmonary artery, the stridor and respiratory distress gradually disappeared. We assume that the occurrence of pulmonary artery sling in distal 18q deletion is not so exceptional but rather it may be missed during routine echocardiographic evaluation as exact echocardiographic imaging of vascular anomalies can be challenging in some patients. In the presented case, echocardiography was the primary and helpful technique for diagnosis of the pulmonary artery sling, which was suspected since the left pulmonary artery was not visualized in the expected location in parasternal short axis view [8]. The definitive diagnosis was made using computed tomography angiocardiography, which is preferred to magnetic resonance imaging due to better visualization of lung parenchyma and airways, higher spatial resolution, and faster scanning with lower requirement of sedation [9–12]. Moreover, computed tomography data can be also used for 3D-printed anatomical model fabrication, which is, nowadays, the highest level of the imaging of congenital heart defects [13,14].

Most individuals with 18q- have terminal deletions, which are associated with neurological problems. The overall level of intellectual disability appears to be mild in patients with deletions distal to 18q21.33 and severe in patients with deletions proximal to 18q21.31 [15]. The presented patient with deletion proximal to 18q21 suffers from severe forms of neurological complications, such as intellectual disability, impaired cognitive, motor, emotional, and communication abilities. New molecular cytogenetic techniques capable of assessment of submicroscopic chromosomal aberrations could provide important insights into the mechanisms underlying intellectual disability in distinct genetic disorders [16].

4. Conclusion

The presented case documents, to our knowledge, the first reported finding of pulmonary artery sling in patient diagnosed with distal 18q deletion. Due to increased risk of congenital cardiac anomaly in the individuals with 18q aberrations, a thorough cardiovascular examination including ultrasound examination by a specialist in pediatric cardiology is recommended. It is likely that the occurrence of pulmonary artery sling in the individuals with distal 18q- is not so exceptional but rather it may be missed during routine echocardiographic evaluation. If a presence of vascular anomalies is suspected, the diagnosis should be confirmed using computed tomography angiography.

Disclosure of Interest

The authors report no conflicts of interest.

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