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Hypoplastic Left Heart Syndrome in a Fetus who died with Marden-Walker Syndrome: A Case Report

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ABSTRACT

This is a case of Marden-Walker syndrome and hypoplastic left heart syndrome. Hypoplastic left heart syndrome accounts for 1.5% to 3.8% of all congenital heart diseases, while Marden-Walker syndrome is a rare genetic disease in which atrial septal defect and ventricular septal defect are the most probable congenital heart diseases. During a diagnostic autopsy I found hypoplastic left heart syndrome in a case of Marden-Walker syndrome. This case is about a single male fetus who was miscarried at 27 weeks of gestation. The cadaver was sent to my laboratory to undergo a diagnostic perinatal autopsy the mother was a 26 years old woman and the father was a 33 years old man who were not consanguineous. The fetus' head and neck presented with edema, telecanthus, small nose, depressed nasal root, anteverted nostril, small jaw (asymmetric micrognathia), short upper eyelids, upper lip eversion, abnormal ear shape, low set ears, and excess skin folds on neck. In the dissected thorax, there was a water bottle shape heart with hypoplastic left heart syndrome. A deletion at some parts of 21q22 is detected in both of hypoplastic left heart syndrome and Marden-Walker syndrome. Although a mutation in PIEZO2 has also been mentioned as a cause of the latter syndrome, this case report can be verification for the etiologic role of 21q22 deletion.

Key words: Marden-Walker Syndrome, Hypoplastic Left Heart Syndrome, Congenital Heart Diseases

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INTRODUCTION

Marden-walker syndrome (MWS) is a rare genetic disease which is caused by mutations in PIEZO2 [1]. Since 1999, at least 30 cases of Marden-Walker had been reported [2]. Aside from PIEZO2 mutation, a 2.84 Mb deletion has also been reported at 21q22.11 in a patient clinically diagnosed with marden–walker syndrome [3], which is an autosomal recessive trait in some families [4]. Although there may be no apparent renal or cardiovascular involvement [5, 6], congenital heart disease is known as a clinical finding of this syndrome [7]. Atrial septal defect (ASD) and ventricular septal defect (VSD) are the most common congenital heart disease seen in it [2,8]. This is a case of hypoplastic left heart syndrome (HLHS) in a 27 weeks old male fetus, as a consequence of MWS, found out during perinatal autopsy.

CASE PRESENTATION

A 27 weeks old male fetus, who had succumbed to intrauterine fetal death (IUFD), was excised from vaginal canal. His mother and father were 26 and 33 years old respectively and they were not consanguineous. This was the mother's first conception, during which screening tests and sonography results were normal. The mother did not present any diabetes, hypertension, urinary tract infection, or spotting during pregnancy. There was no known genetic disease in the parents' families. In the 27th week of gestation no fetal movement was felt and sonography report indicated IUFD.

AUTOPSY FINDINGS

The cadaver was a male fetus who had a crown-heel height of 35.5 cm, and a crown–rump of 23.5

cm. The lower limb was 12cm and his head circumference was 26 cm. There was no bruise but some maceration, especially on the trunk. +1 edema and +1 cyanosis were present. The head and neck examination revealed telecanthus, small nose, depressed nasal root, anteverted nostril, small jaw (asymmetric micrognathia), and short upper eyelids on the face (Fig.1), as well as upper lip eversion. In the lateral view of the face abnormal ear shape, low set ears and excess skin folds on neck were seen (Fig.2).

Figure 1: Telecanthus, small nose, depressed nasal root, anteverted nostril, small jaw (asymmetric micrognathia), upper lip eversion, and short upper eyelids were seen

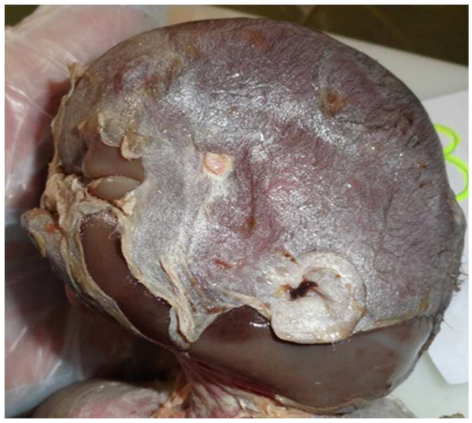

Figure 2: Abnormal ear shape, low set ears, and excess skin folds on neck were prominent

There were no significant findings on the trunk. Extremity findings included arachnodactyly in the fingers, club feet and sandal toes in the feet (Fig.3). The placenta was discoid and 17x11x6 cm. Its membranes were intact. The umbilical cord was 35 cm in the length and 1 cm in the diameter. It had 3 vessels in its cut surface.

In dissection, the thoracic wall was not atrophic and had no emphysema. The diaphragm was intact in the right and the left sides. There was bilateral bloody hydrothorax, with no sign of pneumothorax. The thymus was 2x1x0.7 cm. The heart was water bottle shaped, and measured 4x3.5x2.5 cm (Fig.4). The left ventricle wall was thick (Fig.5), and the other cardiac malformations were normally aligned great arteries without a common atrioventricular junction, underdevelopment of the left heart with significant hypoplasia of the left ventricle including hypoplasia of the aortic and mitral valves, and hypoplasia of the ascending aorta and aortic arch, schematically shown in the figure.6[9]. The lungs were hypoplastic and airless. Each of them was 3x2.5x1.5 cm and they weighed 9 gr. There were normal esophagus and trachea.

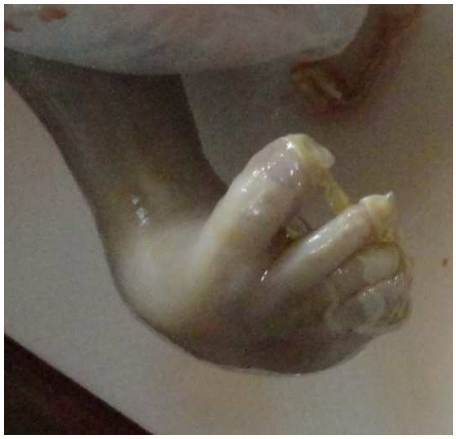

Figure 3: Club feet and sandal Toes were found

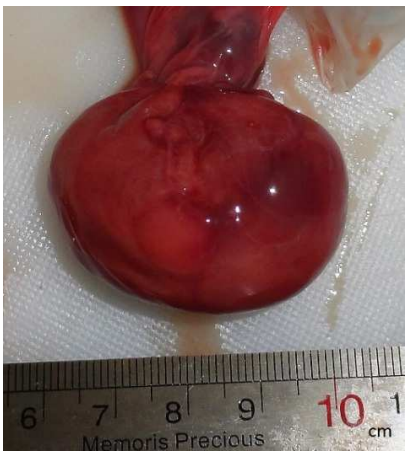

Figure 4: The water bottle shaped heart was seen

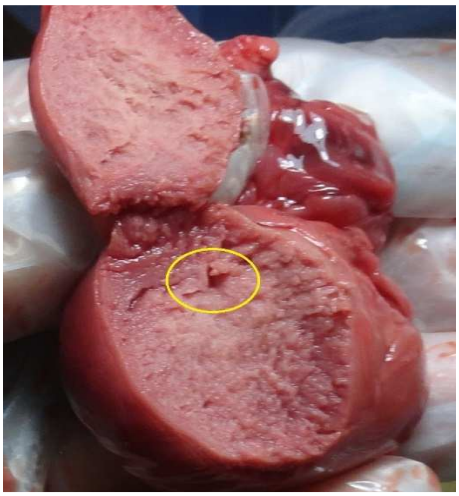

Figure 5: An underdeveloped left ventricle (in the yellow circle) was discovered in the heart section

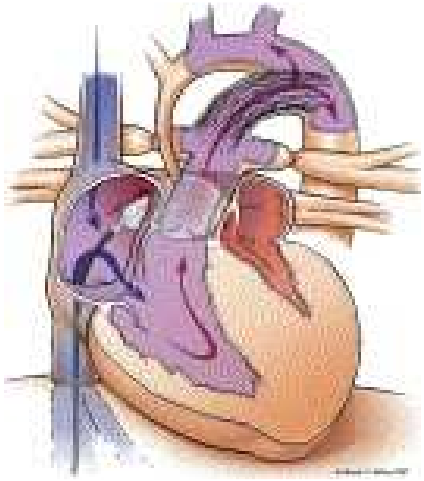

Figure 6: [9] Cardiac malformations are schematically shown, with normally aligned great arteries without a common atrioventricular junction, under development of the left heart with significant hypoplasia of the left ventricle including hypoplasia of the aortic and mitral valves, and hypoplasia of the ascending aorta and aortic arch.

Abdominal cavity was filled with a bloody ascites fluid. The intestinal rotation was normal. The liver was 6x4x2.5 cm with an autolytic consistency. The stomach was 2.2x1.5x0.5 cm and its wall thickness was 0.2 cm. The spleen was bean shape and 2x1.4x0.6 cm. The pancreas was 2.5 cm in the length and 0.4 cm in the diameter. Each ureter had a normal path and a diameter of 0.2 cm. Each kidney was 2.5x1.5c1.3 cm. In cross section, the medulla was hemorrhagic and the cortical thickness was 0.1 cm. Each adrenal gland was 2x1.8x1.3 cm with hemorrhage in its medulla. The bladder wad 1.5 cm in diameter and had a wall thickness of 0.2 cm. It contained bloody urine. Each testicle was 0.8x0.4x0.3 cm and they were both located in the abdominal cavity. The external genitalia and anus were normal. Intracranial elements were autolyzed but if intact, one or some elements of hypoplastic corpus callosum, cerebellar vermis hypoplasia, and enlarged cisterna magna [10] would have been present.

DISCUSSION

HLHS is the eighth most common cardiac defect accounting for 1.5% [11] to 3.8% [12] of all congenital heart diseases (CHD). It is associated with extracardiac anomalies in the 15–28% of cases, in the setting of chromosomal anomalies, mendelian disorders, and organ defects. There has been also, a report on a syndromic female newborn with HLHS and terminal 21q22.3 deletion [13]. Some other case reports also argue that del 21q22 should be added to the list of chromosomal imbalances associated with HLHS [14].

Although ASD and VSD are the most common kinds of the CHD that were reported [2,8] with MWS, this case represents the presence of HLHS with MWS. This can be a document reinforcing the postulate that 21q22.11 deletion [3] is a cause of MWS and the accompaniment of HLHS with 21q22 deletion [14].

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