

CASE REPORT

Moebius syndrome associated with hypoplastic left heart syndrome

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Abstract: The authors present a newborn infant from the second pregnancy whose mother experienced influenza A infection in the first trimester. The birth was at term without complications, but 11 hours later, cyanosis and tachypnoe developed. The final diagnosis was hypoplastic left heart syndrome with simultaneous Moebius Syndrome. In conclusion the authors indicate that echocardiography is of great importance in the diagnosis of this complex heart disease as it helps to analyze in detail the options in the diagnosis of rare Moebius Syndrome (Fig. 2, Ref. 8). Full Text (Free, PDF) www.bmj.sk.

Key words: Moebius syndrome, hypoplastic left heart syndrome.

Congenital Facial Diplegia (Moebius Syndrome) was described for the first time in 1888. Its occurrence is rare, until now it has been reported in approximately 180 cases. Hypoplastic Left Heart Syndrome includes a variety of congenital heart anomalies associated with disturbed development of the aorta, aortal valve, left ventricle, mitral valve and left atrium to varying degrees (1, 2). Simultaneous occurrence of Moebius Syndrome and Hypoplastic Left Heart Syndrome has not yet been described in literature.

Case report

Patient is a child from second pregnancy, and the mother experienced influenza A infection during the first trimester. The birth was a spontaneous delivery in the 41st week of gestation. The newborn baby was female with birth weight of 3420g and length of 52cm. Green amniotic fluid was observed. The Apgar score was 7, 8 and 9 respectively. At the age of 11 hours the child started to become cyanotic, tachypnoic and febrile. The heart rate was 140 per minute. On the precordium a blowing systolic murmur was noted. A second heart sound was accentuated. Pulses were diminished in all extremities.

EKG showed sinus rhythm with electric axis on the right, low voltage over left precordium. Hypertrophy of the right ventricle was present. High peak P wave showed an enlargement of the right atrium. In the chest X-ray, the cardiac silhouette was

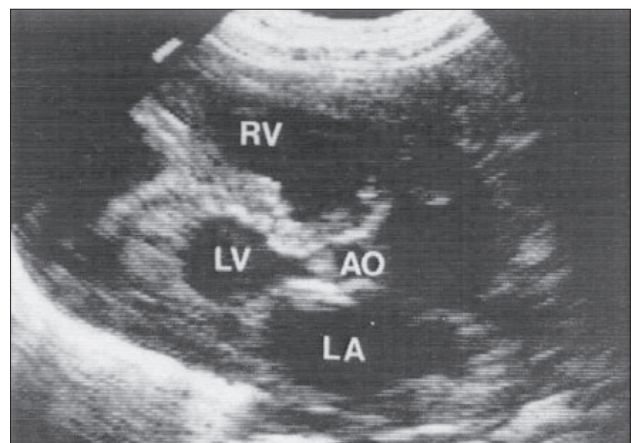


Fig. 1. Echocardiography finding.



Fig. 2. Facial stigmatization.

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globular with an enlargement of the right margin, which meant an enlargement of the right atrium. Pulmonary vascular markings were increased.

Echocardiography illustrated an enlargement of the right atrium, right ventricle and pulmonary artery and hypoplastic left heart and aorta. Coronary arteries were supplied retrogradely through hypoplastic aorta ascendens. The left atrium was small and the interatrial septum was bulging to the right. The ductus arteriosus was wide and enabled the flow of blood from the pulmonary artery into the aorta (Fig. 1).

The child was somatically stigmatized with facial asymmetry and paresis of the facial nerve. She was not blinking with her right eyelid. Her right ear was rudimentary and left ear was normal. The palate was gothic and dysphagia was present (Fig. 2).

Cytogenetic examination excluded chromosomal aberration. Chromosomal analysis of the lymphocytes of peripheral blood confirmed 46, XX karyotype. Neurological examination revealed severe paresis of nerves VII, IX, X, XI, XII on the right side, and a diffused hypotonic syndrome.

Ophthalmologic investigation showed asymmetry of the eye, bigger eyeball, keratitis and lagophthalmus, with normal fundus findings.

On the basis of the clinical and auxiliary findings, the diagnosis of Moebius Syndrome with Hypoplastic Left Heart Syndrome was made.

At the age of 76 days, the child died with signs of cardiopulmonary failure. Autopsy findings confirmed the hypoplastic left heart syndrome and described facial anomalies. Other organs were without congenital macroscopic and microscopic abnormalities.

Discussion

Moebius Syndrome (Congenital Facial Diplegia Syndrome) occurs sporadically. Families in which more than one member has been affected by these diseases have also been described. Autosomal dominant, autosomal recessive and multifactorial inheritance were also considered. In the majority of cases the occurrence is sporadic with unclear causes. In recent years, early embryonal disruption was more implicated. In literature, cases of children with Moebius syndrome were described in mothers who were taking Misoprolol during the first trimester of pregnancy, which is a synthetic analogue of prostaglandins (3).

The syndrome is characterized by a masked face, trismus, drinking and speaking disturbances, strabismus and ptosis. Neurological examinations reveal paresis in one or more of the cranial nerves (nerves VII, VI, III, XII, V, IV, IX). Very often there could be conspicuous microphthalmia, epicanthal folds, decreased lacrimation, lagophthalmus and nystagmus. Ear anomalies and hearing defects can also occur (4). In one third of all patients, uni- and bilateral pes varus are present.

Additional findings may include various hand anomalies associated with homolateral aplasia of the pectoralis muscle (Poland Syndrome) (5, 6). Simultaneous occurrence of congenital arthrogryposis multiplex was also described (4).

Clinical signs (facial symptomatology) are already noticeable after birth. In the newly born and early infancy there usually could be a feeding difficulty with the danger of aspiration. Approximately 90 % of children survive to normal age. In some children with Moebius Syndrome, inherited heart diseases can be detected. In our patient, in addition to Moebius Syndrome, severe complex heart disease – Hypoplastic Left Heart Syndrome – was diagnosed. The prevalence of this syndrome varies between 0.05 and 0.25 in 1000 live-born babies. Hypoplastic Left Heart Syndrome accounts for 1.5 % of the whole group of inherited heart diseases. Some cases of Hypoplastic Left Heart Syndrome were described in relatives. That is why authors of some studies suggest autosomal recessive inheritance (7). Other authors suggest multifactorial inheritance and there is a 0.5 % risk of the occurrence of Hypoplastic Left Heart Syndrome in relatives (8). Although Hypoplastic Left Heart Syndrome occurs together with other extracardial malformations in 10 % of cases (6), the association of Hypoplastic Left Heart Syndrome and Moebius Syndrome has not been described so far in literature.

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Received November 20, 2008.

Accepted March 6, 2009.