Asplenia and polysplenia syndromes with abnormalities of lateralisation in a sibship

J Zlotogora and Elian

From the Department of Pediatrics, Hasharon Hospital, Petah-Tiqva, and Tel Aviv University Medical School, Israel

SUMMARY

The family presented here the first child had asplenia syndrome with cor bicornucarum, transposition of the great vessels, pulmonary stenosis, and anomalies of pulmonary venous drainage and biliary atresia. The second child had polysplenia syndrome with double outlet right ventricle, transposition of the great vessels, pulmonary stenosis, and anomalies of pulmonary venous drainage and biliary atresia. Both children had situs inversus. The possibility that these syndromes, namely asplenia and polysplenia, are different manifestations of a similar defect in the normal asymmetrical development of inner organs is discussed.

CASE 1

A girl was born at term in 1971 after a normal pregnancy and delivery. She was small for gestational age and weighed 2300 g. At the age of 5 days she was referred to the Cardiac Center and a grade 3/6 systolic murmur was heard along the left sternal border. At cardiac catheterisation, transposition of the great vessels with pulmonary stenosis and anomalies of drainage of the great vessels were demonstrated. The child failed to thrive and died at 3 months of age following an episode of septicaemia. The necropsy findings were as follows: cor bicornucarum, transposition of the great vessels with pulmonary stenosis, and anomalies of the pulmonary venous system and drainage of the great vessels with anomalies of the liver. The right lung, being on the right side, was smaller and appeared abnormal. The liver, stomach, and spleen were normal.

CASE 2

In 1978 a 2300 g boy was born to the same family after a normal pregnancy and delivery. At the age of 3 months, a grade 3/6 systolic murmur was heard along the left sternal border. Cardiac catheterisation revealed complete transposition of the great vessels, pulmonary stenosis, and anomalies of pulmonary drainage. At necropsy, the heart sounds were heard at the right side of the sternum, but no murmurs were heard at the left side. The liver, spleen, and stomach were normal. The anomalies of pulmonary drainage and subsequent penile arteries were demonstrated. The right lung, being on the right side, was smaller and appeared abnormal. The left lung, being on the left side, was normal.

REFERENCES

Hasharon Hospital, Petah-Tiqva, and Tel Aviv University Medical School, Israel

Journal of Medical Genetics, 1981, 18, 301-302

Received for publication 12 July 1980

A girl was born at term in 1971 after a normal pregnancy and delivery. She was small for gestational age and weighed 2300 g. At the age of 5 days she was referred to the Cardiac Center and a grade 3/6 systolic murmur was heard along the left sternal border. At cardiac catheterisation, transposition of the great vessels with pulmonary stenosis and anomalies of drainage of the great vessels were demonstrated. The child failed to thrive and died at 3 months of age following an episode of septicaemia. The necropsy findings were as follows: cor bicornucarum, transposition of the great vessels with pulmonary stenosis, and anomalies of the pulmonary venous system and drainage of the great vessels with anomalies of the liver. The right lung, being on the right side, was smaller and appeared abnormal. The liver, stomach, and spleen were normal.

CASE 2

In 1978 a 2300 g boy was born to the same family after a normal pregnancy and delivery. At the age of 3 months, a grade 3/6 systolic murmur was heard along the left sternal border. Cardiac catheterisation revealed complete transposition of the great vessels, pulmonary stenosis, and anomalies of pulmonary drainage. At necropsy, the heart sounds were heard at the right side of the sternum, but no murmurs were heard at the left side. The liver, spleen, and stomach were normal. The anomalies of pulmonary drainage and subsequent penile arteries were demonstrated. The right lung, being on the right side, was smaller and appeared abnormal. The left lung, being on the left side, was normal.

REFERENCES

Hasharon Hospital, Petah-Tiqva, and Tel Aviv University Medical School, Israel

Journal of Medical Genetics, 1981, 18, 301-302

Received for publication 12 July 1980
seen on repeated peripheral blood smear examinations. The chromosomal karyotype was normal. Obstructive jaundice was noted at the age of 3 weeks and the results of laboratory investigations suggested the presence of biliary atresia. Laparotomy at 2 months of age confirmed the intrahepatic atresia and the complete situs inversus. It also revealed a predouodenal portal vein and multiple spleens. A Roux-en-Y portoenterostomy was performed, but because of inadequate function a second portoenterostomy was necessary. The child died some hours after the operation.

The necropsy showed the complete situs inversus and the multiple malformations characteristic of the polysplenia syndrome. Both lungs were bilobed and symmetrical and there was dextrocardia with atrial and ventricular septal defects, transposition of the great vessels with double outlet of the right ventricle, and pulmonary stenosis. In addition, anomalous pulmonary venous return was also demonstrated, each pulmonary vein being connected to the corresponding lung. The inferior vena cava terminated in the azygous vein. The liver was very enlarged and bilobed, occupying the upper abdomen almost completely. On the right side, the spleen was enlarged and bilobed, and in addition four accessory spleens were present in the isthmus.

Discussion

Both asplenia and polysplenia syndromes affect all the major internal organs and are closely related. From the clinical standpoint these two disorders can be separated. In the asplenia syndrome the cardiovascular malformations tend to be more complex and severe, there is an increase in severe infection, Howell-Jolly bodies are seen in peripheral blood smears, and the spleen is absent on the scan, while the liver is central. In polysplenia the associated defects are usually less severe or may even be absent; an increased incidence of biliary atresia has been reported in these patients. The diagnosis can be made by demonstration of multiple spleens on the abdominal scan.

The occurrence of splenic anomalies is rare in sibships. Only eight families have been reported in which asplenia was present in more than one sib, and one family with two affected sibs with polysplenia has been reported.

In the family reported here and the family described by Polhemus and Schaefer, one child had asplenia and another polysplenia. All the four affected children demonstrated very similar severe cardiovascular defects. The close similarity of the anomalies found in four children of two families seems to be more than a coincidence, and the question arises whether the asplenia and polysplenia syndromes present in these children are secondary to a similar disturbance, most probably congenital. This disturbance leading to a failure of normal asymmetrical development could result in a bilateral symmetry of the right or the left side.

The assumption of a primary defect of laterality led Smith to classify both syndromes as 'laterality anomalies'; laterality abnormal with bilateral right sidedness for asplenia, and laterality abnormal with bilateral left sidedness for polysplenia.

The mode of inheritance of both syndromes remains unclear. The possibility of a multifactorial origin of autosomal transmission has been suggested.

In all instances when a sib is affected by one of these syndromes, the other sibs have to be examined not only for the same syndrome, but also for the other splenic syndrome. It seems that the best results can be obtained by abdominal scanning.

References


Requests for reprints to Dr Ezra Elian, Department of Pediatrics, Hasharon Hospital, Petah-Tiqwa, Israel.
Asplenia and polysplenia syndromes with abnormalities of lateralisation in a sibship.
J Zlotogora and E Elian

doi: 10.1136/jmg.18.4.301

Updated information and services can be found at:
http://jmg.bmj.com/content/18/4/301

**Email alerting service**
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

**Notes**

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/