Unusual association of diseases/symptoms

Fanconi's anaemia and cerebrovascular anomaly

M P Mahato,¹ D B Sharma,¹ Rashmi Shukla²¹Department of Paediatrics, ASCOMS and Hospital, Jammu, India;²Genetic Unit, Department of Paediatrics, All India Institute of Medical Sciences, New Delhi, India**Correspondence to** Dr Mathura Prasad Mahato, dr_mathuraprasad@yahoo.com**Summary**

Fanconi's anaemia (FA) is a paradigm of congenital anomalies including almost all systems. But FA with cerebrovascular anomalies is rare. The authors report a case of FA with hypoplasia of the posterior cerebral circulation including the basilar artery, bilateral vertebral arteries, posterior cerebral and posterior communicating arteries. The diagnosis of FA was established on the basis of induced chromosomal breakage study with mitomycin C.

BACKGROUND

Fanconi's anaemia (FA) is associated with multisystem congenital defects but the association of FA with cerebrovascular abnormalities is rare. Published literatures of association between FA and cerebrovascular abnormalities or FA with Moyamoya disease are very few. Hence, we decided to write it up.

CASE PRESENTATION

A 12-year-old boy was admitted with presenting complaint of progressive pallor, bleeding gums, headache and vomiting since 15 days. There was no history of fever,

convulsion or neurological deficit. His three elder sisters and one elder brother are normal.

On examination he was well-oriented and interactive. He was short stature (125.0 cm) and underweight (17.0 kg). Both height and weight were less than fifth percentile. Hypertension (150/90 mm Hg) was found with heart rate 96/min. He was pale with petechiae over lower limbs and bleeding gums. He had café au lait spots over face and trunk. He had radially curved right forearm with absent thumb. There was no lymphadenopathy and liver and spleen were not enlarged. There was no neurological deficit. Genitalia were normal. Other systems were normal.

Figure 1 MRI angiogram of brain showing hypoplasia of basilar artery, posterior cerebral and posterior communicating arteries.

Figure 2 Peripheral blood lymphocytes culture induced with mitomycin C showing high frequency of chromosomal breakage.

INVESTIGATIONS

His haematological profile revealed anaemia (Hb-4.0 g/dl), thrombocytopaenia (platelet count-35 000/mm³), reticulocytopaenia (reticulocyte count-0.2% of RBC) and total leucocytes count was 5800/mm³. Bone marrow was hypocellular. Skeletal roentgenic survey revealed hypoplastic right radius and ulna with absence of all phalanges of the thumb. Ultrasound of abdomen revealed hypoplastic left kidney. Echocardiography was normal.

MRI angiogram of brain revealed hypoplasia of basilar artery, posterior cerebral and posterior communicating arteries. (figure 1).

Cytogenetic studies performed on peripheral blood lymphocytes cultures induced with mitomycin C¹ revealed high frequency of chromosomal breakage (figure 2). Cultures without mitomycin C induction showed no chromosomal breakage.

DIFFERENTIAL DIAGNOSIS

- ▶ Familial pancytopenia
- ▶ Dyskeratosis congenita.

TREATMENT

Fresh blood transfusion was given to correct anaemia and thrombocytopaenia. Antihypertensive, hydralazine was added to treat hypertension.

Bone marrow transplantation could not be done due to unavailability of suitable donor.

At present, the child is on low-dose steroid (prednisolone) and antihypertensive drug, hydralazine.

OUTCOME AND FOLLOW-UP

Since past 18 months, the child is doing well with low dose of prednisolone and antihypertensive, hydralazine but getting frequent respiratory tract infection.

DISCUSSION

FA is named after a Swiss paediatrician who described in 1927 three brothers who had pancytopenia with other birth defects.^{1 2} Since then FA associated with multisystem defects, that is, marrow failure, skeletal anomalies, renal anomalies, pulmonary dysgenesis, cardiovascular malformations, gonadal hypoplasia, etc has been reported. The diagnosis of FA is based on characteristic chromosomal breaks following clastogenic stress.^{3 4} FA with cerebrovascular anomalies is rare and hence the present case is of academic importance. FA with moyamoya pattern cerebrovascular malformation has been reported by Cohen *et al*⁵ in a 10-year-old boy. Palvokis *et al*⁶ reported two patients with FA and Moyamoya disease taken from a database composed of 434 FA. Crisitiene M Rocha *et al* reported a 14-year-old boy with FA and agenesis of internal carotid artery.^{4 6}

Learning points

- ▶ FA, a paradigm of congenital anomalies, may be associated with cerebrovascular malformation.
- ▶ MRI angiogram should be done for any case of FA who develops symptoms or signs pertaining to central nervous system.
- ▶ Cytogenetic studies performed on peripheral blood lymphocytes culture induced with mitomycin C reveals chromosomal breakage to establish the diagnosis of FA.
- ▶ Other cytogenetic abnormalities in FA include defective DNA repair, increased susceptibility of haematopoietic cells to oxidant stress and decreased cell survival.

Competing interests None.

Patient consent Obtained.

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Please cite this article as follows (you will need to access the article online to obtain the date of publication).

Mahato MP, Sharma DB, Shukla R. Fanconi's anaemia and cerebrovascular anomaly. *BMJ Case Reports* 2011;10.1136/bcr.01.2010.2680, date of publication

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