Thrombocytopaenia with absent radius (not radii)

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DESCRIPTION

A 13-year-old boy presented to the outpatient department with incidentally detected thrombocytopaenia. Historically, the patient had no bleeding symptoms or cow milk intolerance. Physical examination revealed hypoplastic thumbs in both hands and thinned-out right forearm (figures 1–3). X-ray of both forearms showed absent radius on the right side with bilateral hypoplastic thumbs (figure 4A, B). Systemic examination otherwise was essentially normal. His haematological parameters revealed haemoglobin of 12.5 g/dL, total leucocytes 4900/ μ L and platelet counts 46 000/ μ L. Bone marrow examination revealed a reduction in thrombopoietic element (hypomegakaryocytic thrombocytopaenia) with preservation of other haematopoietic elements. Chromosomal breakage studies performed were negative.

Our patient has two unique features. One, unlike conventional cases of thrombocytopaenia with absent radius (TAR), our patient has a fully developed radius in the uninvolved side with bilateral hypoplastic thumbs. Second, the patient's elder brother was diagnosed with Fanconi's anaemia at 10 years of age with normal parents/grandparents.

TAR syndrome is characterised by normal thumbs with absent bilateral radius. It is an autosomal recessively inherited disorder with microdeletions in 1q21.1.¹ Various associations reported by Greenhalg *et al* include lower limb involvement and lactose intolerance (particularly cow's milk) with 47% cases of each, renal anomalies (27%) and congenital heart diseases in 15% cases.² Various studies evaluated thrombocytopaenia of the TAR syndrome; its suggested mechanisms include defective c-Mpl signalling and dysmegakaryocytopoiesis

Figure 2 The right upper limb showing thinned-out forearm due to absent radius and hypoplastic thumb.

characterised by cells blocked at an early stage of differentiation.³ TAR needs to be differentiated from other syndromes such as thalidomide embryopathy, Holt-Oram syndrome, Robert syndrome and Fanconi's anaemia.

Figure 1 Palmar aspect of the right upper limb showing thinned-out forearm due to absent radius and hypoplastic thumb.

Figure 3 Comparison of the right (pathological) and left forearms and hands, both hands have hypoplastic thumbs with the left forearm having an intact well-developed radius.

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Figure 4 (A) X-ray of the right arm, forearm and hand showing absent radius and hypoplastic thumb. (B) X-ray of the left forearm and hand showing hypoplastic thumb and an intact well-developed radius as compared with the right forearm.

Learning points

- ▶ Thrombocytopaenia with absent radius is a rare autosomal recessive congenital disorder with bilateral absent radius.
- ▶ Congenital thrombocytopaenia is associated with a number of syndromes associated with limb anomalies.
- ▶ It is very important to perform thorough general examination for congenital anomalies in young patients with thrombocytopaenia to help in diagnosis of congenital thrombocytopaenia syndromes.

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