

A case of Cows' milk allergy in the syndrome of thrombocytopenia with absent radius

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Introduction

Thrombocytopenia with absent radii (TAR) is a rare congenital disorder with hypomegakaryocytic thrombocytopenia and bilateral radial aplasia.¹ They presents in early infancy ranging from mild changes to marked limb shortening. About 50% of the patients with TAR syndrome have cow's milk protein allergy.² Here we report a 2 year old girl with TAR syndrome presented with marked gastrointestinal disturbance and clinical evidence of cows' milk allergy.

Case report

She was the first child born to non-consanguineous parents following uneventful antenatal period. She had an obvious forearm deformity at birth. (Figure 1)



Figure 1



Figure 2

There were bilateral radial club hands with radial aplasia, (Figure 2) whereas the rest of the skeleton was normal.

She had thrombocytopenia (30 x 10³) with normal other haematological indices during neonatal period and has had marked thrombocytopenia during early infancy without any bleeding manifestations which was improved in subsequent full blood counts. Rest of her full blood count parameters was normal. She thrived well with exclusive breastfeeding for six months. There were no concerns with the commencement of complementary feeding. However, since the introduction of cow's milk at 14 months, she developed episodic abdominal distension and blood stained stools with failure to thrive.

Cow's milk products were introduced for the first time around 14 months. The stools contained mucus and blood, but repeated stool culture showed no gastrointestinal pathogens. There were no features of inflammatory bowel disease. Direct relationship with the cow's milk product and the severity of symptoms were observed. Decision was made to withhold the cow's milk products and her gastrointestinal symptoms improved markedly. As a diagnostic trial cow's milk was reintroduced after 2 weeks and she developed similar GI symptoms again. None of these episodes precipitated haematological relapses. She never had urticaria, vomiting or edema suggestive of Ig E mediated allergy. Child was managed without cow's milk products and she remained asymptomatic thereafter.

Discussion

In 1969 TAR syndrome was first defined as amegakaryocytic thrombocytopenia and absent radii.³ The molecular basis of TAR syndrome has recently been identified as a microdeletion on Chromosome 1.⁴ Though this child didn't have, Lower limb skeletal abnormalities are also associated with TAR syndrome with lesser frequency. Unlike congenital amegakaryocytic thrombocytopenia (CAMT), platelet count tends to improve in TAR syndrome after 1 year. In this case she had persistently

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Case Reports

normal platelet count since early infancy.⁵

Cow's milk protein allergy is a known association of TAR syndrome. It is well known that haematological relapses occurs in patients with TAR when they exposes to cow's milk products, this was not evident in this child. Cow's milk protein allergy occurs both in IgE mediated and non IgE mediated ways. This patient had features suggestive of non IgE mediated allergy. Diagnosis of non IgE mediated cow's milk protein allergy is challenging. Food challenge is a one method of diagnosing it. In this child there was a clear relationship with the GI symptoms and the cow's milk products consumption.²

The mainstay of treatment for any food allergy is the appropriate identification and elimination of foods responsible for food hypersensitivity reactions.⁶ At the same time it is important to prevent nutritional deficiencies due to the dietary modification.

Since it's a common association of TAR syndrome, any such children presenting with gastrointestinal symptoms should be evaluated for possible cow's milk protein allergy.

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