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CASE REPORT

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# Thrombocytopenia with absent radii syndrome with a congenital absence of patella: A case report

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## ABSTRACT

Thrombocytopenia-absent Radius (TAR) syndrome is manifested by a bilateral absence of the radius with the presence of the thumbs, thrombocytopenia, and a number of other signs such as congenital heart abnormalities, facial dysmorphia, and kidney defects. Prevalence is estimated to range from 0.5 to 1/100,000. In all patients, a deletion of chromosome 1q21.1 was found. Management is multidisciplinary; it can include continuous surveillance of hemorrhagic syndrome, transfusion by platelet and/or sometimes globular units, and orthopedic treatment. The prevention of bleeding in the first years of life remains essential, as the prognosis of this syndrome is more critical during this period. We report a case of TAR syndrome with a congenital absence of patella in a 6-year-old child.

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## Introduction

Thrombocytopenia-absent radius (TAR) syndrome is a very rare congenital condition inherited in an autosomal recessive pattern [1]. Prevalence is estimated to range from 0.5 to 1/100,000 [2]. This syndrome is characterized by bilateral absence of the radius with the presence of thumbs, thrombocytopenia, and a number of other signs, such as congenital heart defects, facial dysmorphia, and renal malformations. The etiology and mode of transmission remain unclear. All patients have a deletion of chromosome 1q21 [3,4].

We report the case of a Thrombocytopenia with Absent radii syndrome with a congenital absence of patella, diagnosed on the basis of clinical examination and characteristics observed during cross-sectional imaging magnetic resonance imaging (MRI) and X-rays of the forearm, knees, and pelvis [3,4].

## Case Report

We report the case of a 6-year-old child who was seen in the emergency room for abdominal pain and diarrhea after ingesting cow's milk.

The initial examination revealed an erythematous rash on the buttocks with no change in general condition or fever.

Physical examination revealed that the child was small for his age and that both arms were hyperflexed at the elbow, with radial deviation of the hands and bilateral thumbs (Fig. 1), present since birth, along with a low platelet count (68,000/ul).

The child had a history of fluctuating thrombocytopenia consistent with TAR syndrome, with lower counts during infancy and progressive improvement with age.

There were no other associated signs, notably no neurological deficit, otological sign, or cervical adenopathy.

An X-ray of the upper limbs revealed the absence of the radius on both sides (Fig. 2).

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**Figure 1.** Clinical appearance of a radial club hand.

Given this radiological, clinical, and biological picture, TAR syndrome was suspected.

Since patients with TAR syndrome often have abnormalities in other organs and systems, detailed additional tests were performed.

The echocardiogram, renal ultrasound, and spinal ultrasound were normal.

A pelvic X-ray was also performed, which showed no abnormalities, but the knee X-ray revealed bilateral absence of the patella. An MRI of the knees was then requested, revealing patellar dysplasia classified as DUJOUR 3 with bilateral quadriceps tendon hypertrophy, and damage to the medial retinaculum on the left side (Figs. 3–6).

Since the patient is currently 6 years old, platelet counts in TAR syndrome typically improve with age, reducing hemorrhagic risk after early childhood. Ongoing follow-up in this patient includes: periodic hematologic monitoring of platelet levels, orthopedic evaluation for the lower limb, and functional development.

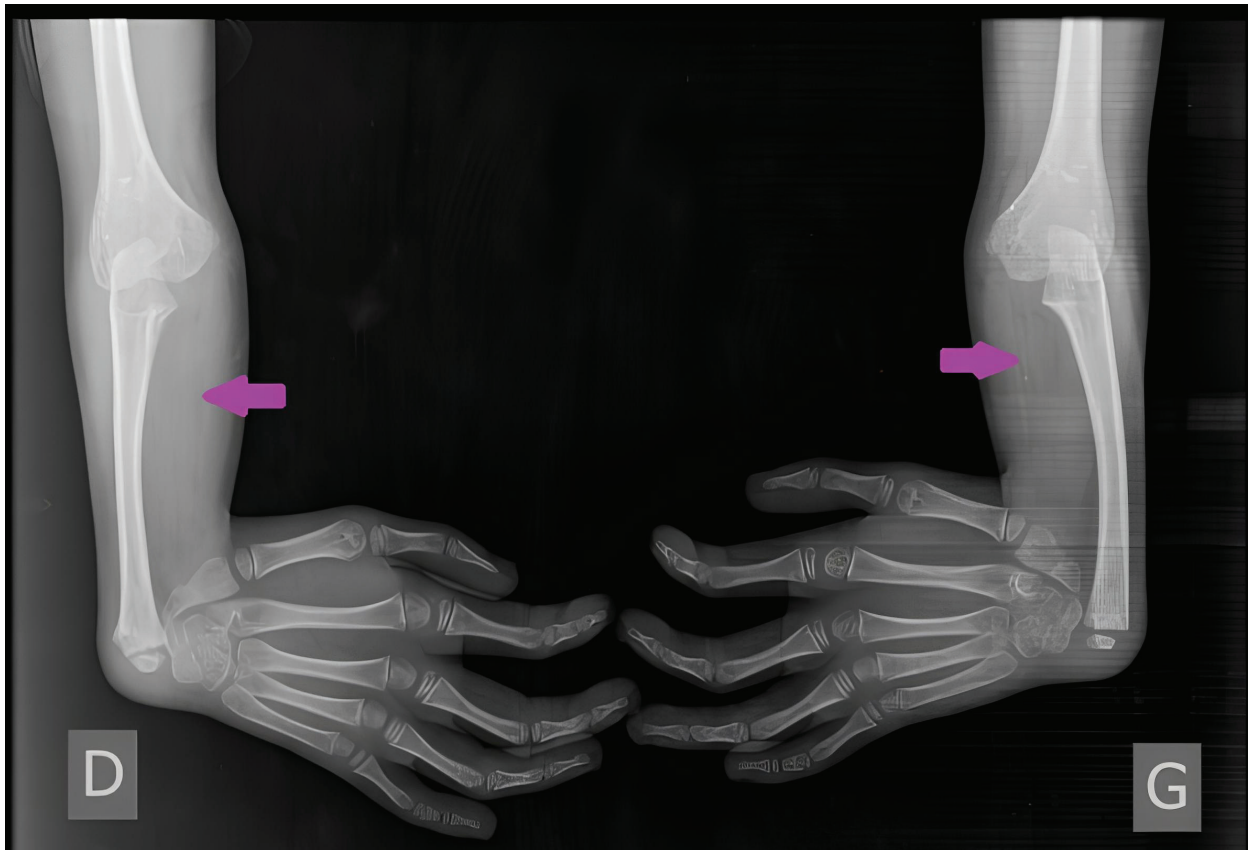



**Figure 2.** X-ray of the forearm showing the absence of the radius on both sides (purple arrow).

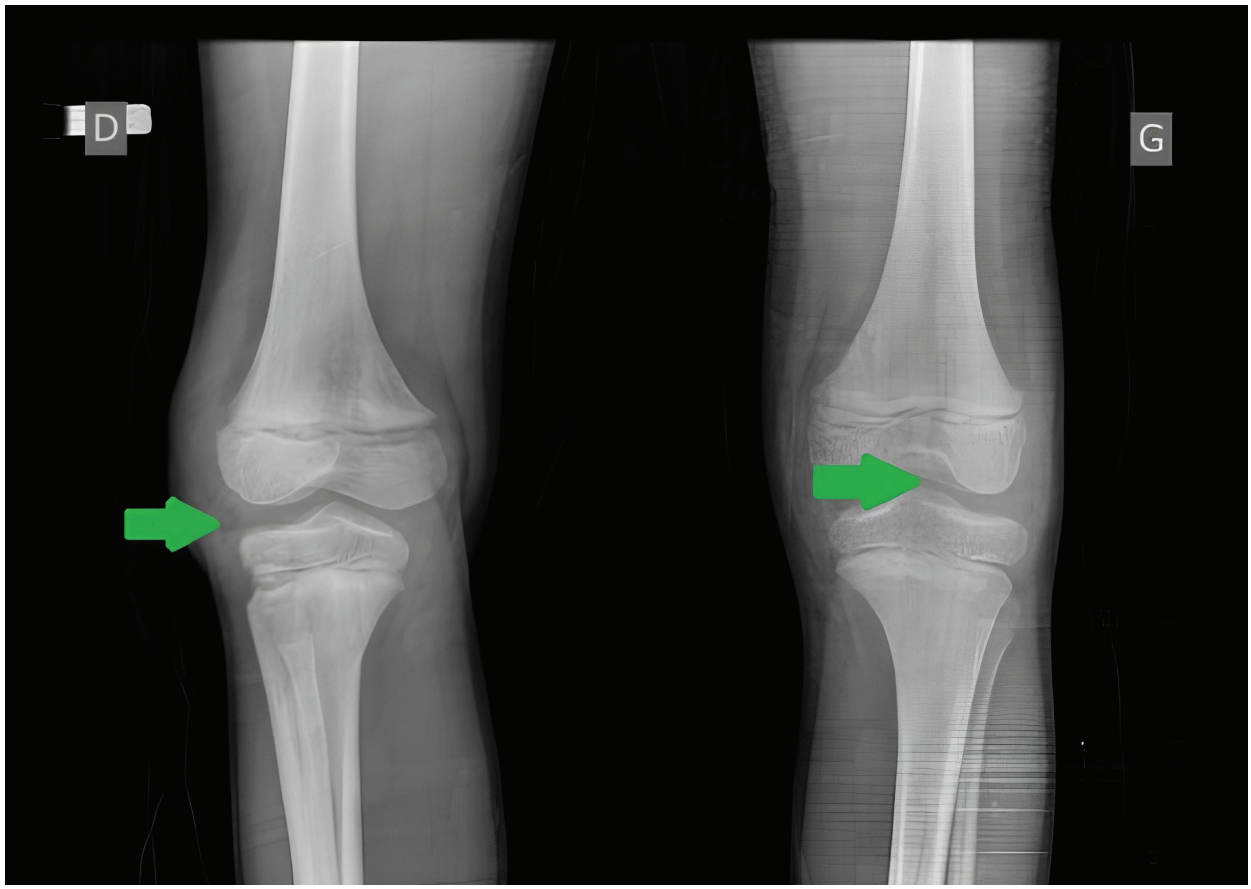



**Figure 3.** Frontal X-ray of the knee showing the absence of the patella (Green arrow).

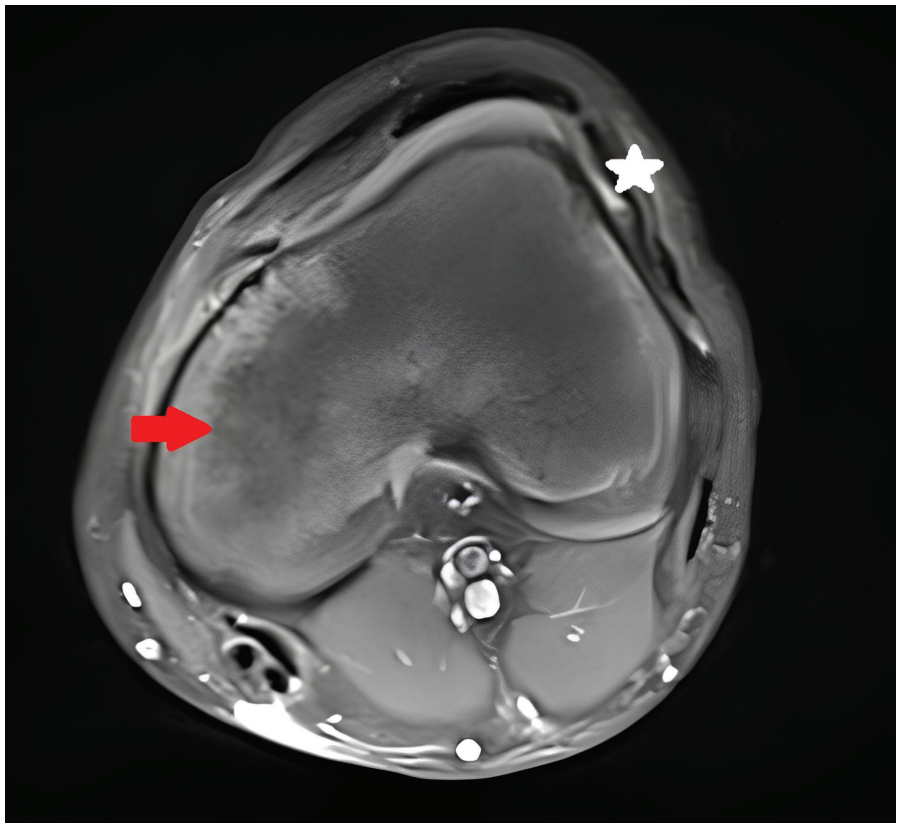


**Figure 4.** X-ray of the pelvis without abnormalities.

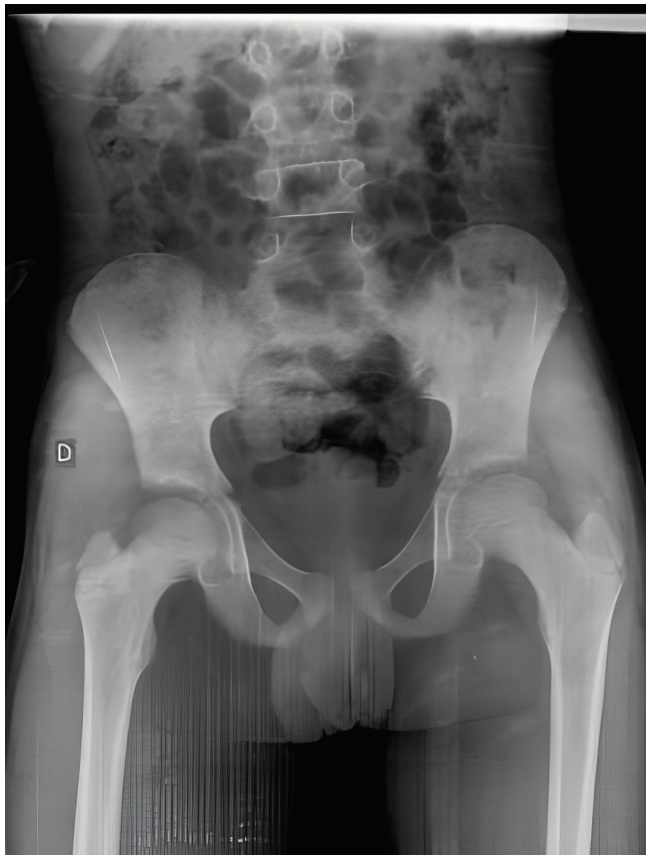



**Figure 5.** Axial DP FAT MRI sequence showing left trochlear dysplasia, classified as Dujour 3 (Red arrow) associated with thickening of the median retinaculum (white star).

The preliminary diagnosis, based on clinical, biological, and radiological criteria, favored a TAR syndrome with a congenital absence of patella.

## Discussion

TAR syndrome follows an autosomal recessive inheritance pattern that typically involves compound inheritance: a microdeletion at chromosome 1q21.1 on one allele and a hypomorphic variant (most commonly a specific SNP) in the *RBM8A* gene on the other allele [5]. This combination leads to reduced expression of the RBM8A protein (Y14), which is responsible for the clinical phenotype [6].

Diagnosis of the syndrome can occur prenatally by ultrasound and platelet count via fetal blood sampling via cordocentesis [7].

At birth, the absence of radii and the presence of thrombocytopenia, with or without other manifestations of TAR syndrome, are considered hallmarks of a positive diagnosis [8].

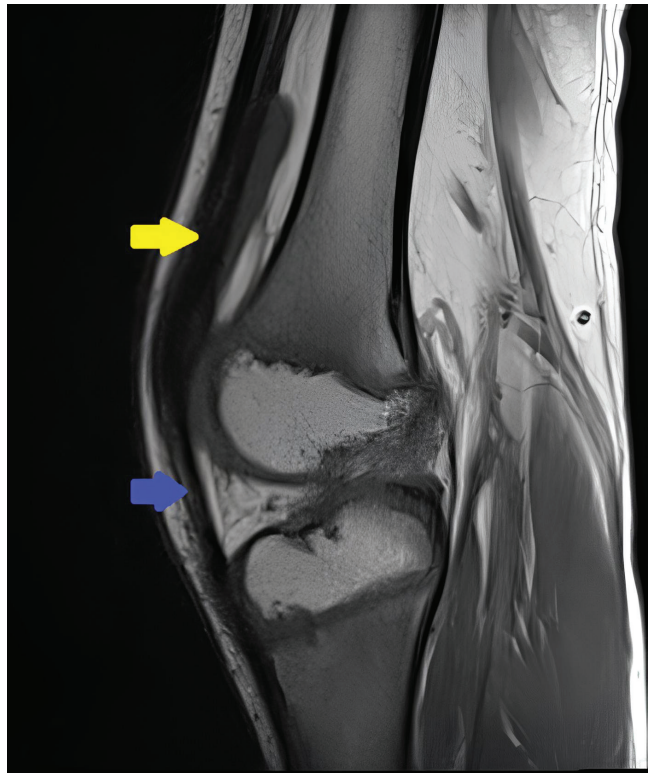


**Figure 6.** T1 MRI sequence in sagittal section, showing patellar agenesis (blue arrow) associated with quadriceps tendon hypertrophy (yellow arrow).

The thrombocytopenia that develops in TAR syndrome is Hypomegakaryocytic Thrombocytopenia (HMT), platelet hypoproduction caused by insufficient numbers of megakaryocytes, the precursors to platelets, in the bone marrow [8].

The etiology of the HMT in these patients is hematopoietic stem cells, which do not appropriately respond to thrombopoietin and impaired maturation of the megakaryocyte progenitor cells in the bone marrow [9].

Thrombocytopenia may be present at birth among those with TAR syndrome, with 95% of those with the condition developing significant thrombocytopenia within 4 months of birth [9]. Platelet counts appear to increase with age and can appear normal in later childhood/adulthood [10].

Moreover, there is a possibility of exacerbation of the underlying thrombocytopenia by Cow's Milk Protein Allergy. The mechanism of the exacerbation is reported to be direct immunoglobulin E immune-mediated or secondary to increased gastrointestinal bleeding bleeding due to loss of coagulation proteins [10].

It is important that healthcare professionals treating those with TAR syndrome are aware of its impact on platelet count and act accordingly when performing surgical procedures [11].

Patients with TAR syndrome have absent radii with preserved fingers and thumbs bilaterally. These patients tend to have a contracture of the joint, which causes flexion and radial deviation of their hands, resulting in the hands often being at right angles to the forearms [12].

The syndrome frequently results in hypoplasia of the muscles and soft tissues in the shoulders and arms. Shoulder abnormalities associated with TAR syndrome may include absent or hypoplastic scapula, glenoid fossa, clavicle, or acromion [13].

TAR syndrome has been known to be associated with multiple cardiac, gastrointestinal, renal, skeletal, and hematologic anomalies [14]. Although the rarity of TAR syndrome limits our ability to explore its epidemiology through the use of large datasets, one clinical genetic study explored the phenotypes of 34 patients with confirmed TAR syndrome. They found all the cases had thrombocytopenia and bilateral radial aplasia; concurrently, 47% also had lower limb malformations, patellar agenesis specifically is a less common finding that warrants awareness from orthopedic surgeons, 23% had renal anomalies, and 15% had cardiac anomalies [15].

Renal and genitourinary abnormalities may be associated with TAR syndrome. Case reports have described patients with TAR syndrome as having horseshoe kidneys, penoscrotal transposition, bilateral hypoplastic kidneys with decreased renal function, and crossed fused renal ectopia [16].

Treatment is multidisciplinary and may include continuous monitoring of the hemorrhagic syndrome, transfusion of platelet concentrates and/or sometimes red blood cell concentrates, and orthopedic treatment with splints and corrective orthoses [17].

The prognosis for TAR syndrome is more critical during the first 2 years of life, a period during which children develop several episodes of potentially serious hemorrhagic syndromes such as cerebral hemorrhage [17].

## Conclusion

TAR syndrome is an exceedingly rare genetic condition with a clear presentation. It is associated with increased morbidity and mortality among children diagnosed with it. Potentially severe when diagnosed within the first few weeks of life, the condition can be effectively treated, and its consequences reduced.

## Conflits of interest

The authors declare no conflict of interest (s).

## Author contributions

All the authors contributed to the conduct of this research work. The authors have read and approved the final version of the manuscript.

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