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Case Report

Thrombocytopenia and Absent Radius Syndrome (Tar Syndrome) with Hypoplastic Left Kidney: A Rare Case Report

Sanjay P[1], Patil M M[2], Dhamangaonkar B R[3], Pattanshetti R C[4], Patil S V[5]

[1]Post-graduate Student, [2]Senior Resident, [3]Professor, [4]Professor & Head, [5]Assistant Professor;
Department of Radio-Diagnosis, BLDE University, Shri B.M.Patil Medical College Bijapur Karnataka India.

Corresponding Author: Sanjay P

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ABSTRACT

Thrombocytopenia and absent radius syndrome (TAR) is a rare genetic disorder which is inherited as an autosomal recessive trait. This disorder is characterized by low levels of platelets in blood (thrombocytopenia) resulting in potentially severe bleeding episodes primarily during infancy. Other characteristic findings include bilateral radial aplasia. Other abnormalities may also be present such as defects of hands, structural malfunctions of the heart and kidneys.

Keywords: Thrombocytopenia, absent radius, hypoplastic left kidney, autosomal recessive.

INTRODUCTION

Thrombocytopenia and absent radius syndrome (TAR) is a rare genetic disorder which is inherited as an autosomal recessive trait. This disorder is characterized by low levels of platelets in blood (thrombocytopenia) resulting in potentially severe bleeding episodes primarily during infancy. Other characteristic findings include bilateral radial aplasia. Other abnormalities may also be present such as defects of hands, structural malfunctions of the heart and kidneys.

The prevalence of TAR syndrome is estimated at 1:200,000-1:100,000.

CASE PRESENTATION

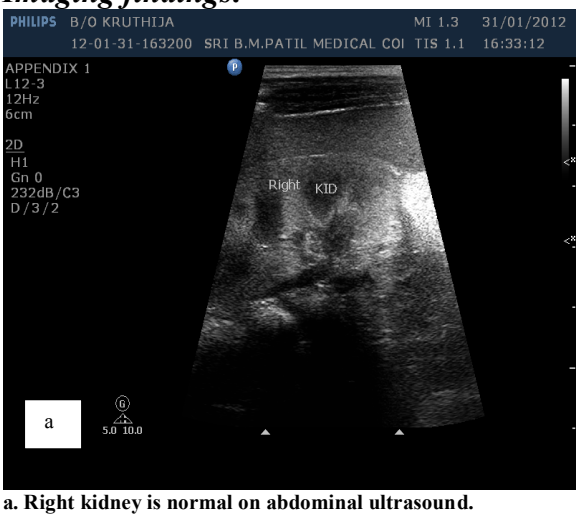
History: A one and half month old female child born to a couple of II degree consanguineous marriage, presented with abnormality of both upper limbs and umbilical swelling since birth and was referred to the RadioDiagnosis department for imaging.

The baby was a product of a full term pregnancy and delivered through normal vaginal delivery and baby cried immediately after delivery. There was no history of birth trauma and no family history of similar illness.

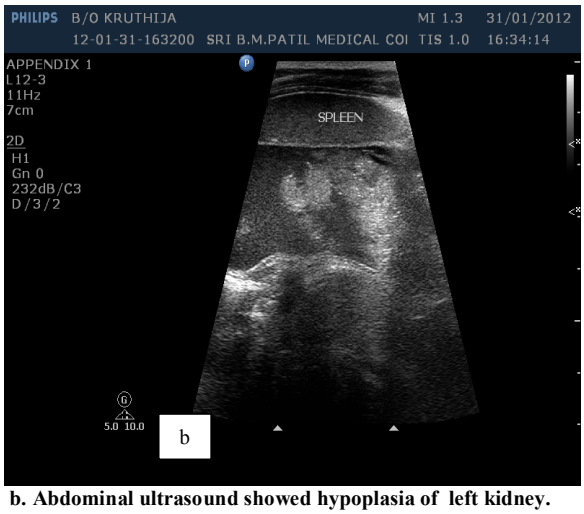
Examination: On musculoskeletal examination, both forearm were shortened with associated bowing and fixed flexion at the elbow and wrist joint.

Lab Findings: Platelet count : 48,000/ mm^3.

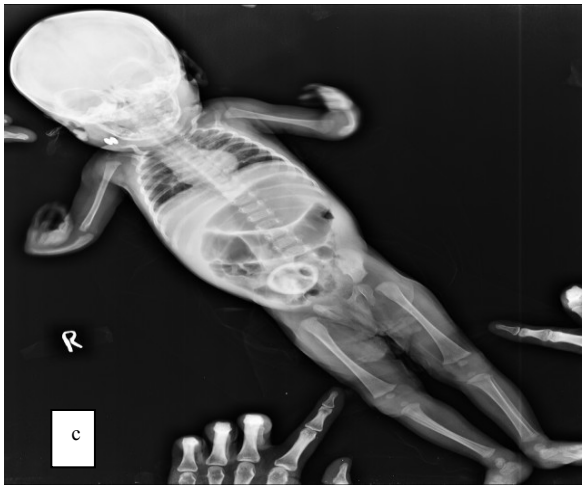
Imaging findings:



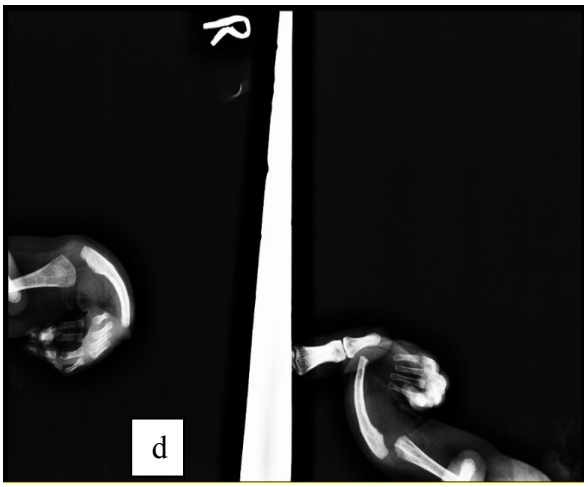
a. Right kidney is normal on abdominal ultrasound.



b. Abdominal ultrasound showed hypoplasia of left kidney.



c. Plain radiograph of both the upper limbs showed congenital absence of radius.



d. Bowing of the ulna and flexion deformity of the elbow and wrist joint.

DISCUSSION

TAR syndrome is a rare genetic disorder characterized by low platelet level and radial aplasia.[1]

A case of a one and half month old female with TAR syndrome was presented to highlight the radiological features

Thrombocytopenia, which may be transient, is seen in 100% of cases diagnosed with TAR syndrome.[2]

The upper limb abnormalities range from isolated absent radii with normal thumbs, as seen in our case.

Lower extremity anomalies occur in 46% of patients but are usually less severe than those of the upper limbs. These abnormalities include hip dislocation, femoral torsion, tibia torsion and deformity of the knee.[3] None of these were seen in our case except hypoplastic left kidney.

The etiology of radial aplasia in TAR syndrome is a primary failure of chondrogenesis and not a disorder of vasculogenesis as in other disorders with absent radii.[4]

In an attempt to understand the genetic basis of TAR syndrome, Klopocki et al. reported that TAR syndrome has a complex pattern of inheritance associated with a common interstitial microdeletion of 200 kb on chromosome 1q21.1 and an additional, as yet unknown, modifier.[5]

Houeijeh et al. mentioned that the identification of the 1q21.1 deletion allows for confirmation of the TAR syndrome diagnosis, particularly in patients with atypical phenotypes, and it also allows for accurate genetic counselling, especially when it occurs de novo.[6]

The genomic structure of the 1q21.1 breakpoint regions is extremely complex, with at least four large segmental duplication blocks ranging in size from 270 kb to 2.2 Mb. Within 1q21.1 there are two areas where a deletion can be found: the TAR area for the TAR syndrome and the distal area for other anomalies. The 1q21.1 deletion syndrome will commonly be found in the distal area, but an overlap with the TAR area is possible.[7]

Eye abnormalities are seen in 26% of individuals with 1q21.1 microdeletion and may include strabismus, chorioretinal and iris colobomas, microphthalmia, hypermetropia, Duane anomaly, and congenital cataract.[8]

CONCLUSION

TAR syndrome is a rare genetic disorder characterized by low platelet level and radial aplasia. A case of a one and half month old female with TAR syndrome was presented to highlight the radiological features. This patient had congenital absence of radius with bowing of the ulna and flexion deformity of the elbow and wrist joint with hypoplasia of left kidney. The patient was transfused with platelet concentrate having presented with low platelet count, and she improved thereafter.

REFERENCES

1. Hall JG, Levin J, Kuhn JP, Ottenheimer EJ, van Berkum KA, McKusick VA: Thrombocytopenia with absent radius (TAR). Medicine 1969, 48:411–440.
2. Omran et al.: Thrombocytopenia and absent radii (TAR) syndrome associated with bilateral congenital cataract: a case report. Journal of Medical Case Reports 2012 6:168.
3. Ballmaier M, Schulze H, Strauss G, Cherkaoui K, Wittner N, Lynen S, Wolters S, Bogenberger J, Welte K: Thrombopoietin in patients with congenital thrombocytopenia and absent radii: elevated serum levels, normal receptor expression, but defective reactivity to thrombopoietin. Blood 1997, 90:612–619.
4. Furman GI, Martin GI: Visual diagnosis casebook: thrombocytopenia with absent radius. J Perinatol 1988, 8:166–168.
5. Klopocki E, Schulze H, Strauss G, Ott CE, Hall J, Trotier F, Fleischhauer S, Greenhalgh L, Newbury-Ecob RA, Neumann LM, Habenicht R, König R, Seemanova E, Megarbane A, Ropers HH, Ullmann R, Horn D, Mundlos S: Complex inheritance pattern resembling autosomal recessive inheritance involving a microdeletion in thrombocytopenia-absent radius syndrome. Am J Hum Genet 2007, 80:232–240.

6. Martinovic J, Escande F, Devisme L, Bisiaux S, Chaffiotte C, Baux M, Kerckaert JP, Holder-Espinasse M, Manouvrier-Hanu S: Thrombocytopenia-absent radius (TAR) syndrome: a clinical genetic series of 14 further cases. Impact of the associated 1q21.1 deletion on the genetic counselling. Eur J Med Genet 2011, 54:e471–e477.
7. Redon R, Ishikawa S, Fitch KR, Feuk L, Perry GH, Andrews TD, Fiegler H, Shapero MH, Carson AR, Chen W, Cho EK, Dallaire S, Freeman JL, González JR, Gratacòs M, Huang J, Kalaitzopoulos D, Komura D, MacDonald JR, Marshall CR, Mei R, Montgomery L, Nishimura K, Okamura K, Shen F, Somerville MJ, Tchinda J, Valsesia A, Woodwark C, Yang F, et al: Global variation in copy number in the human genome. Nature 2006,444:444–454.
8. Mefford HC, Sharp AJ, Baker C, Itsara A, Jiang Z, Buysse K, Huang S, Maloney VK, Crolla JA, Baralle D, Collins A, Mercer C, Norga K, de Ravel T, Devriendt K, Bongers EM, de Leeuw N, Reardon W, Gimelli S, Bena F, Hennekam RC, Male A, Gaunt L, Clayton-Smith J, Simonic I, Park SM, Mehta SG, Nik-Zainal S, Woods CG, Firth HV, et al: Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. N Engl J Med 2008, 359:1685–1699.

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