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Absent Radii, Radial Clubhand, Hip Dysplasia and Left Coxofemoral Dislocation in A 3-Year-Old Boy

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Abstract Absent radii a rare skeletal condition marked by the complete failure of the formation of bilateral radius during pregnancy, often associated with radial clubhand and multiple systemic abnormalities. We present a case of a 3 years old boy with absent radii, bilateral radial clubhand, left hip dysplasia and left coxofemoral dislocation associated with pulmonary hypoplasia, right renal hypoplasia and bilateral cryptorchidism. This report describes a rare case and highlights the importance of early control to direct diagnosis, to be aware of the large number of complications that can occur in other organ systems and quickly care for associated abnormalities and find strategies to preserve the functionality.

Keywords: *Absent radii, bilateral radial clubhand, congenital longitudinal deficiencies of the radius hip dislocation*

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1. Introduction

Congenital longitudinal deficiencies of the radius are various anomalies such as partial agenesis of the bone to its total absence which can present with a spectrum of upper limb deficiencies [1] and is often associated with abnormalities in other organ systems, such as cardiac and renal, and so requires a comprehensive medical evaluation [2]. Absent radii is a rare skeletal condition marked by the complete failure in the development of structures that form the radius bone in both forearms [3], this rare condition is often associated with radial clubhand that is a descriptive term that was first mentioned by Jean-Louis Petit in 1733 who reported a case of a male newborn with bilateral absent radii, also the absent radii could be related with multiple systemic abnormalities. Herein we present a case of a 3 years old boy with absent radii, bilateral radial clubhand, hip dysplasia associated with pulmonary hypoplasia, right renal hypoplasia and bilateral cryptorchidism, due to the characteristics of the case, TAR syndrome vs Fanconi anemia is suspected, and we defined surgery priorities in order; the most important is correction of left acetabular and femoral alteration; next improving range of motion and preserve function of clamp in hands. All this evaluates the viability of possible surgical interventions according to the patient's bone characteristics. This report describes a rare case and highlights the importance of early control to direct diagnosis and find strategies that help preserve the functionality of upper and limbs and try to correct lower limb deformity.

2. Case Presentation

A 3 years-old kid who was brought for consultation to study multiple malformations in his upper and lower limbs diagnosed during prenatal check-ups. The patient was born of a 17 year old mother product of first pregnancy at 39 weeks, weight 2.950 gr and size 50 cms, the mother presented maternal hypothyroidism in substitution with levothyroxine, complete STORCH negative, denies consumption of psychoactive substances, or other medications, denies paternal consanguinity and the mother also refers paternal history of epilepsy without pharmacological management, denies others. The patient has a medical record that shows pulmonary hypoplasia, right renal hypoplasia and bilateral cryptorchidism treated by bilateral orchidopexy 20/03/2021.

On physical examination with multiple congenital malformations; low implantation of the ear pinnae, micrognathia, bilateral deltoid atrophy and shoulder girdle muscle atrophy, absent radii, bilateral radial clubhand (Figure 1) that retains clamping function, elbows with arcs of mobility that begin at 45 degrees of flexion up to 135 degrees of passive flexion, hip and acetabular dysplasia with abduction limitation, shortening of the left femur (Figure 2), lameness due to shortening of the left lower extremity.

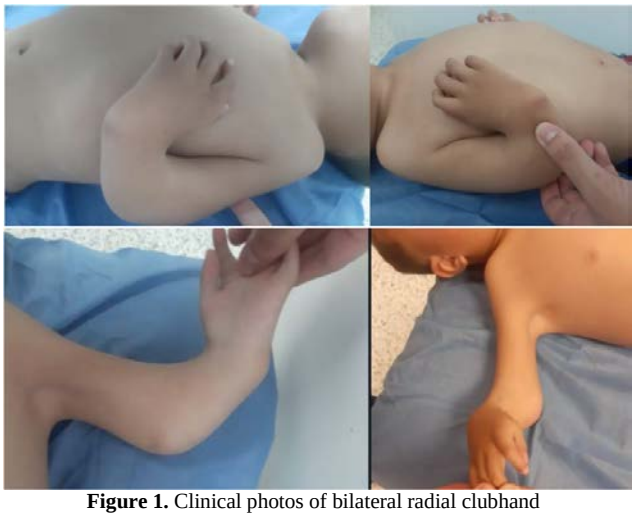

Figure 1. Clinical photos of bilateral radial clubhand

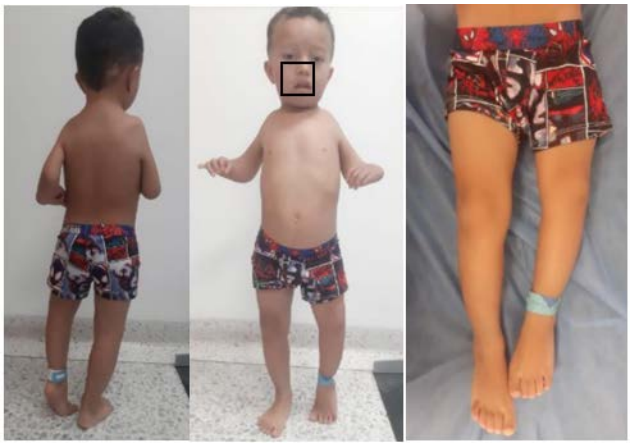

Figure 2. Clinical photos of shortening of the left femur

Comparative radiographs of the forearms and hands showed total absence of bilateral radius, severe radial deviation of the hand and wrist (Heikel IV).

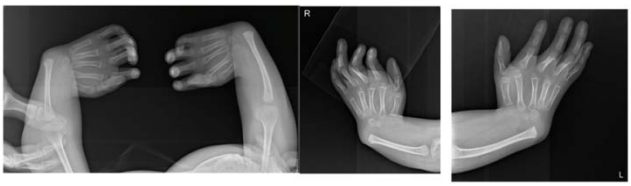

Figure 3. Comparative forearm and hand X-ray

And radiographs of the hip showed left hip dysplasia and left coxofemoral dislocation

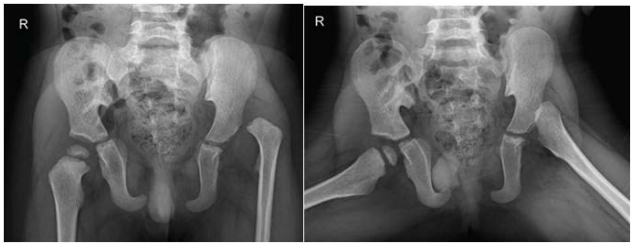

Figure 4. Hip X-ray

He also had a complete blood count that reported severe thrombocytopenia and pancytopenia as a newborn and tended to normalize around two years of age.

Based on the clinical history, physical examination and imaging findings, we made a diagnosis of thrombocytopenia absent radius syndrome (TAR) vs Fanconi anemia taking into account the findings of absent radii, bilateral radial clubhand, left hip dysplasia and left coxofemoral dislocation

3. Discussion

Congenital longitudinal deficiencies of the radius are various anomalies such as partial agenesis of the bone to its total absence. Birch-Jensen reported 73 cases of congenital longitudinal deficiencies in a sample of 4 million people [4], thus defining a frequency of 1 in 100,000 newborns.; bilateral in approximately half of the cases and the prevalence has been reported to be slightly higher in boys, at 3:2 [5]. Radial clubhand that is a descriptive term that was first mentioned by Jean-Louis Petit in 1733 who reported a case of a male newborn with bilateral absent radii [6], Kato reviewed 250 cases of radial clubhand and is considered in the medical community as the first attempt at a comprehensive study of this condition [7].

It has been proposed that the congenital absence of the radius is due to suppression of the first accessory ray described by Gegenbauer which is composed of the radius and large navicular and multiangular bones of the carpus, the first metacarpal and the two phalanges of the thumb, this would also explain the usual relationship between these disorders and the absence of the thumb [8]. Everything indicates that the most probable cause of radial clubhand is injury to the apical ectoderm or deep mesenchymal tissues on the anterior surface of the primordium of the developing upper limb [9].

Environmental factors that favor the appearance of this type of deficiency must also be taken into account, such as; exposure to thalidomide, radiation and viral infections that must act before the sixth week of gestation because by this time the upper limb has completed its development [9].

It is important to explore conditions associated with radial absence (Table 1), because at the moment in which the primordium of the upper limb develops, several organs develop simultaneously, this also with the aim of identifying a future risk in surgical approach.

The main concerns being blood dyscrasias such as Fanconi anemia defined as an autosomal and X-linked recessive disorder characterized by bone marrow failure, acute myelogenous leukemia, solid tumors, and developmental abnormalities [10] or TAR syndrome that is a rare autosomal recessive disease characterized by hypomegakaryocytic thrombocytopenia and bilateral radial aplasia. Its expression includes skeletal, hematologic, and cardiac system abnormalities [11] with an incidence of 0.42\100 000 population [12] and the presence of cardiac anomalies such as Holt-Oram syndrome, an autosomal dominant disorder which occurs because of mutations in the TBX5 genes. Most notable manifestations include musculoskeletal deformities, predominantly affecting the upper limbs, and congenital heart defects that have also been noted in 75% of patients [13,14].

Our patient has multiple congenital malformations that made us think about TAR syndrome vs Fanconi anemia, this is being studied with genetics, the patient is awaiting surgery to correct defects in his lower limb due to left hip dysplasia and left coxofemoral dislocation, then we will focus on maintaining clamp function of the hands and mobility of his upper limbs.

Table 1. Congenital malformations that may be part of the condition of congenital longitudinal radial deficiency

Hand	Cardiac abnormalities
Thumb	-Atrial septal defect (Holt-Oram syndr
-Absent	-Ventricular septal defect
-Floating	-Coarctation of aorta
-Hypoplastic	-Persistent ductus arteriosus
	-Dextrocardia
Fingers	-Tetralogy of Fallot
-Syndactyly	-Pulmonary stenosis
-Polydactyly	
-Symphalangia	Genitourinary abnormalities
-Triphalangy	-Renal agenesis
	-Kidney hypoplasia
Carpus	-Horseshoe kidney
-Deficiency on the radial side - Absence of scaphoid and trapezius	-Pelvic kidney
-Carpal shock	-Hydronephrosis
	-Urethral valve
Metacarpophalangeal	-Neurogenic bladder
-Excessive hyperextension limited flexion	
	Gastrointestinal abnormalities
Proximal interphalangeal	-Esophageal atresia
-Fixed flexion deformity	-Rectovaginal fistula
	-Imperforate anus
Column	-Inguinal hernia
-Congenital scoliosis	
Hemivertebrae	Lung abnormalities
-Klipple-Feil syndrome	-Agenesis of the upper lobe of the lung
-Sacral agenesis	
-Idiopathic scoliosis	Head abnormalities
	-Cleft lip and palate
Skeleton	-Craniosynostosis
-Hip dislocation	-Hydrocephalus
-Congenital high scapula	-Eye cataract
-Short, arched ulna	-Coloboma
-Radioulnar synostosis (partial absence of the distal radius)	-Ear anomalies
clubfoot	
	Chromosome abnormalities
Sternal abnormalities	-Trisomy 18
-Pectum excavatum	
-Chest in keel	

Note: By Tachdjian. (2001). Pediatric Orthopedics (second Edition). p 206 (3)

There are certain complications, it is a difficult patient to follow up because of his social condition, the follow-up period of this case was short and it has not been possible to continue as desired. However, the main goals in his treatment could be defined from the pediatric orthopedics service.

We consider that additional large-scale studies are needed to help guide this type of patient and that it would benefit from a multidisciplinary assessment.

4. Conclusion

The presentation of this rare clinical case of a patient with absent radii, radial clubhand and multiple congenital malformations is important to highlight the attention of a series of complications that can be improved to reduce and avoid functional consequences in the future.

Our goal in publishing the current case report is to make clinicians aware of the large number of complications that can occur in other organ systems so that they can quickly diagnose and care for any associated abnormalities. Also the patient will need an early multidisciplinary assessment to avoid loss of function and have a better follow up of his disease.

Declaration of conflicts of interest

Each author declares that he/she has no commercial and/or personal association that could have any conflict of interest in connection with this article.

Ethics

As a minor the authorization of his legal representative was necessary so the authors socialized with her an informed consent and she signed to approve the publication of this case report. The information described was shared with her prior to its publication.

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