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Langer Giedion Syndrome with Absence of Bilateral Radial Arteries – A Rare Clinical Association

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ABSTRACT

Langer Giedion syndrome, also known as Trichorhinophalangeal syndrome type 2 (OMIM 150230) is a rare multisystem disorder characterized by distinctive facial features and cartilaginous exostosis. It is a contiguous gene deletion syndrome located on chromosome 8q24.1-q24.13. The characteristic phenotypic features include sparse hair, multiple cone shaped epiphysis, multiple cartilaginous exostoses, bulbous nasal tip, thickened alar cartilage, upturned nares, prominent philtrum, large protruding ears, mild mental retardation and stunting of growth. The authors present a 6 year old boy with classical features of Langer Giedion syndrome with absence of radial artery bilaterally as a rare clinical association which has never been described previously.

Keywords: *Trichorhinophalangeal syndrome type II, exostoses, microdeletion, dysmorphic features, bilateral absence of radial arteries*

INTRODUCTION

The trichorhinophalangeal syndrome (TRPS) was first described in 1966 by Giedion [1]. It affects the tricho (hair), rhino (nose), phalanges (fingers) and has been classified into three types. Trichorhinophalangeal syndrome type 2 (TRPS2) is also known as Langer Giedion syndrome (LGS). It was first described by Andreas Giedion,

a Swiss pediatric radiologist and Leonard O Langer Jr., an American radiologist.

TRPS2 is a true contiguous gene deletion syndrome with deletions in both TRPS1 and EXT1 genes on chromosome 8q24.1-q24.13[2]. It is characterised by a bulbous, pear shaped nose, elongated philtrum, sparse hair, cone shaped epiphysis, multiple exostoses and growth retardation[3]. TRPS2 combines features of Trichorhinophalangeal syndrome type1 along with multiple exostoses[4].

In our case, the bilateral absence of radial artery with thenar hypoplasia is a unique feature which has never been described in association with this syndrome.

CASE REPORT

A 6 year old, male child, presented with failure to thrive and multiple exostoses over the scapulae and knees. He was born of a non-consanguineous marriage, had two elder female siblings who were normal. Child was immunized till date and was developmentally normal.

On examination, his weight and height were below the 3rd percentile for age. Vital parameters were normal. A striking feature was noted, wherein both the radial pulses were absent along with thenar hypoplasia. Central and peripheral pulses in all the limbs were present. Blood pressure measurement in both upper limb and lower limb was normal for age. Other findings included brachycephaly, squint, microtia of right ear, low set ears, hypertelorism, short bulbous nose, deformed ear cartilage, poor dentition, and overlapping of toes(Fig1). Multiple cartilaginous exostoses were seen over the ribs, scapulae and knees (Fig 2). Ophthalmic examination did not reveal any abnormality. Systemic examination including the cardiovascular system was normal.

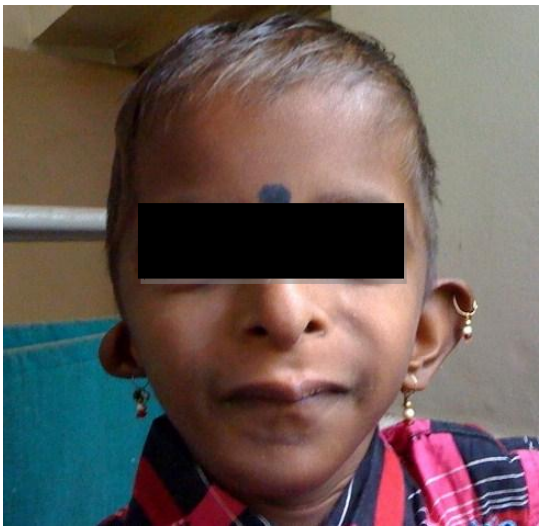

Fig 1 Langer Giedion Syndrome, Note the Sparse Hair, Squint, Bulbous Nose, Microtia of Right ear, low set Ears, Thin Vermillion Border

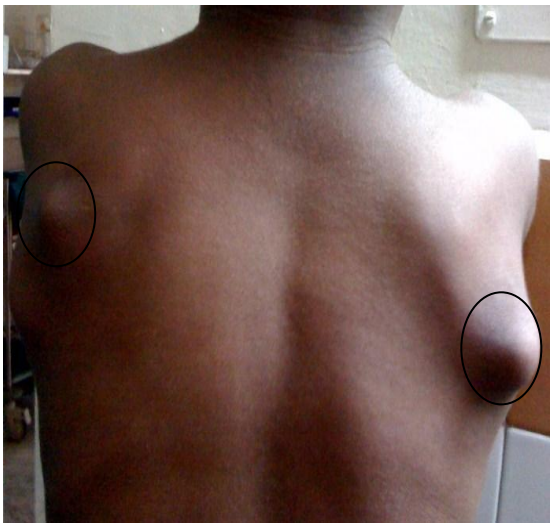

Fig 2. Exostoses seen over the scapular region

Radiograph of the hands revealed delayed bone age, cone shaped epiphysis and multiple exostoses bilaterally. His intelligence quotient was normal.

On colour doppler study, the arteries of the right and left upper limb, that is, the brachial, ulnar and interosseous arteries showed low resistance flow, while the radial artery could not be visualized from the bifurcation on both sides. The right and the left lower limb doppler studies revealed no abnormality. Ultrasonography of the abdomen was normal. Chromosomal analysis in the form of karyotyping was done which was normal.

DISCUSSION

TRPS 2 is a contiguous gene deletion syndrome with deletions in both TRPS1 and EXT1 genes located on chromosome 8q24.1-q24.13. The deletion on the EXT1 gene differentiates it from TRPS1. When larger pieces of 8q are deleted, mental retardation is more likely to occur in TRPS2[5].

TRPS 2 occurs sporadically while some cases of autosomal dominant inheritance have been described in literature[6,7]. It is characterized by craniofacial and skeletal abnormalities such as sparse hair, bulbous nose, long philtrum, thin upper vermillion border, short stature and cone shaped epiphysis[8].

Mutiple cartilaginous exostoses distinguishes TRPS2 from TRPS1. Multiple exostoses may result in pain, limited range of joint movement, and pressure on nerves, blood vessels, the spinal cord, and tissues surrounding the exostoses. Surgical removal of exostoses is indicated in such cases[3].

CONCLUSION

To conclude, our case is an unusual one, as the child presented with a classical LGS phenotype but without a gene deletion in TRPS1 and EXT1 genes(A similar case was presented by Pereza et al,2012)[9]. Another striking feature was the absence of radial arteries bilaterally along with thenar hypoplasia.

REFERENCES

1. Giedion A. Tricho-rhino-phalangeal syndrome. Helv Paediatr Acta. 1966 Nov;21(5):475-85.
2. Carlos A. Bacino, Brendan Lee. Abnormalities of chromosome structure (Deletions and duplications).In : Kleigman RM, Stanton BF, Gemelll JW, Schor NF, Behrman RE,eds. Nelson Textbook of Paediatrics. 19th Ed (Vol 1). Philadelphia : Saunders (Elsevier), 2013: pp. 406 408t.
3. Helen T Shin, Mary Wu Chang.Trichorhinophalangeal Syndrome, Type II (Langer-Giedion Syndome) Dermatology Online Journal (2001)7(2): 8.
4. Piccione, Maria, et al. "Identification of two new mutations in TRPS 1 gene leading to the tricho-rhino-phalangeal syndrome type I and III." *American Journal of Medical Genetics Part A* 149.8 (2009): 1837-1841.
5. Ludecke HJ, Wagner MJ, Nardmann J, et al. Molecular dissection of a contiguous gene syndrome: localization of the genes

involved in the Langer-Giedion syndrome. Hum Mol Genet 1995;4(1):31-6.

6. Murachi S, Nogami H, Oki T, et al. Familial tricho-rhino-phalangeal syndrome Type II. Clin Genet. 1981;19(3):149-55.
7. Shabtai F, Sandowski U, Nissimov R, et al. Familial syndrome with some features of the Langer-Giedion syndrome, and paracentric inversion of chromosome 8, inv 8(q11.23----q21.1). Clin Genet. 1985;27(6):600-5.
8. Langer LO, Krassikoff N, Laxova R, et al. The tricho-rhinophalangeal syndrome with exostoses (or Langer-Giedion syndrome): four additional patients without mental retardation and review of the literature. Am J Med Genet1994; 19:81-111.
9. Pereza N., Severinski S., Ostojic S, et al. Third case of 8q23.3-q24.13 deletion in a patient with Langer-Giedion syndrome phenotype without TRPS1 gene deletion. Am J Med Genet. 2012; 158A:659-663.