Polyarthralgia revealing a trichorhinophalangeal syndrome: A case report and review of the literature

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ABSTRACT

Introduction: Trichorhinophalangeal dysplasia (TRPS) or Langer-Giedion syndrome is a rare genetic disease belonging to the acrodysplasia group. It is an autosomal dominant disorder, characterized by hair growth, nose and phalanges anomalies. Its prevalence is unknown. This syndrome is due to chromosomal microdeletion of variable size in the region 8 q24.12 resulting in the loss of at least two genes: TRPS1 and EXT1. Individuals with TRPS type 1 have a long range of orthopedic problems ranging from hyper laxity to osteochondritis that develop in adolescence but may be occur in infancy resulting in limited mobility and psychological impact. The diagnosis of this dysplasia is based primarily on clinical and radiological features which allows radiology to be the management cornerstone of this syndrome.

Case Report: A 13-year-old girl with polyarthralgia was diagnosed with this syndrome. The genomic study and radiological assessment were in favor of the diagnosis. A medical treatment was administered with a favorable evolution. There were no sign of articular degeneration in the follow up examination. Conclusion: Management of Langer-Giedion syndrome must start with genetic counseling to establish an early diagnosis in order to preserve the functional prognosis. The favorable evolution under medical treatment makes it possible to avoid further surgery in patients with TRPS by proposing a simple radiological monitoring.

Keywords: Polyarthralgia, Radiography, Trichorhinopharyngeal dysplasia, Thymic hyperplasia

INTRODUCTION

Trichorhinophalangeal dysplasia (TRPS) or Langer-Giedion syndrome is a rare genetic disease belonging to the acrodysplasia group. It is an autosomal dominant disorder, characterized by hair growth, nose and phalanges anomalies. This syndrome is due to chromosomal microdeletion of variable size in the region 8 q24.12 resulting in the loss of at least two genes: TRPS1 and EXT1. Individuals with TRPS type 1 have a long range of orthopedic problems ranging from hyper laxity to osteochondritis that develop in adolescence but may be occur in infancy resulting in limited mobility and psychological impact. The clinical and radiological features are major criteria for diagnosis of this dysplasia [1].

CASE REPORT

A 13-year-old girl, who was diagnosed by her school doctor for bone deformities of the hands and feet,
addressed to our unit for follow-up. The medical history of our patient revealed that the child was born of a consanguineous marriage. She is the last of three sibling family. Her 19-year-old brother and his 18-year-old sister had the same bone deformities but they never consulted. On clinical examination, it was found that there was a stature and weight regression (-2 SD), short and sparse hair, and a bulbous nose.

There was a bilateral clinodactyly of the last two phalanges of the middle finger and the ring finger with shortening of the little finger, and at the feet level the clinodactyly affected the second Hallux. The radiographic assessment of the hands revealed a bilateral misalignment of the last two phalanges of the fingers predominant at the level of the middle finger, the ring finger and the little finger. An irregular and curved aspect of the metaphyses of the middle phalanges (inverted V) and epiphyses of conical shape. There was also a last left metacarpal shortening (Figure 1).

The feet X-rays examination had shown a bilateral misalignment of the last phalanx of the Hallux with irregular and curved aspect of the proximal metaphyses of the first phalanx of the two Hallux and a conical deformation of the opposite epiphyses (Figure 2). The pelvis radiographs had shown a bilateral involvement of the femoral head, which was flattened and carried by a short neck, as well as widened and deformed in varus (Figure 3). The set of clinical and radiological abnormalities allowed us to evoke the diagnosis of Trichorhinophalangeal dysplasia Type 1 (TRPS1). This was confirmed by the genetic study which revealed an interstitial chromosomal deletion of 8q24.12.

DISCUSSION

Trichorhinophalangeal dysplasia (TRPS) is a rare hereditary dysplastic syndrome belonging to the group of acrodysplasia. It is a malformation syndrome characterized by a small size, sparse hair, a bulbous nose, conical epiphyses, as well as phalangeal, metacarpal and metatarsal shortening.

Three types of TRPS were described (Table 1), among them Types 1 and 3 are variants of the same entity [2]. The association of intellectual deficit and the presence of numerous abnormalities, particularly multiple exostoses, distinguish Tricho-rhino-phalangeal dysplasia Type 2, also known as Langer-Giedion syndrome.

TRPS is an inherently autosomal dominant disease. The mutation of the gene involved is localized in 8q24.12. Sporadic cases have been reported in Type 2 [1–3].
The diagnosis of TRPS is based on clinical and radiological aspects. Antenatal diagnosis based on DNA is possible if the mutation in the family has been identified [4] which suggest that DNA studies can be interesting in screening for these dysplasias.

Clinically, patients with tricho-rhino-phalangeal dysplasia type 1 have sparse hair, a bulbous pear-shaped nose and a type of hands and feet clinodactyly and brachydactyly. It is also characterized by delicate and fragile nails and a slight staggering delay [4–6].

Trichorhinophalangeal dysplasia type 3 has the same characteristics as type 1, except that stunting and brachydactyly are more severe.

Regarding patients with tricho-rhino-phalangeal type 2 dysplasia, they have in addition to the listed above characters, multiple exostosis, a hyperextension joints anomaly, microcephaly with prominent ears, excess skin, and severe mental retardation [7–9].

On the radiological level, the anomalies observed involve extremities in particular. At there is enlargement, irregularities and curvature of the middle phalanges metaphysis which will take an “inverted V” shape. A reduction of the growth cartilage with a premature epiphyses welding. This metaphyseal asymmetry development is responsible for a clinodactyly of the corresponding phalanx. We observed that the index and middle fingers are usually the most involved, also epiphyses the proximal or distal phalanges. The densification of the epiphysis of the latter is frequent. Another characteristic anomaly of this dysplasia is metacarpal shortening [10, 11].

The pelvic radiographs finds femoral epiphyses deformations. Unilateral involvement is frequent. The femoral head appears to be crushed and deformed with insufficient coverage as it can be observed in the sequelae of the osteochondritis. For some authors, femoral epiphyses deformations are the result of an osteochondritis gone unnoticed [10, 11].

Table 1 [12] describes all the features of the three types of Trichorhinophalangeal dysplasia. The differentials of TRPS includes other syndromes which associates hair growth and structural abnormalities of the nose as well as an osteoarticular involvement: Larsen syndrome, oro - digito - facial syndrome, Coffin - Siris chondrodysplasia and McKusick. Although considered rare, trichorhinophalangeal dysplasia remains the most frequent syndrome to date [4].

Management should begin with genetic counseling, as early diagnosis can better prevent joint damage and preserve functional prognosis.

In case of pain the analgesics for short duration therapies with a radiologic monitoring. Orthopedic surgery is often necessary to maintain joint mobility. It is sometimes necessary to replace a damaged joint, especially in case of hip involvement, with a prosthesis [4]. Plastic surgery can sometimes be necessary to correct the facial dysmorphism in case of aesthetic discomfort.

In our case, the patient was managed by analogics for short duration with radiologic monitoring as well as genetic counseling for the family. To this day, there was no necessity to provide joint replacement surgery.

CONCLUSION

Management of Langer-Giedion syndrome must start with genetic counseling to establish an early diagnosis in order to preserve the functional prognosis. The favorable evolution under medical treatment makes it possible to avoid further surgery in patients with TRPS by proposing a simple radiological monitoring.

REFERENCES


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Author Contributions
Lamiae Bouimetarhan – Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Ayouche Othman – Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
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Guarantor of Submission
The corresponding author is the guarantor of submission.

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Consent Statement
Written informed consent was obtained from the patient for publication of this case report.

Conflict of Interest
Authors declare no conflict of interest.

Data Availability
All relevant data are within the paper and its Supporting Information files.

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