Case report

MR IMAGING OF PACHYDERMOPERIOSTOSIS

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SUMMARY

A case of pachydermoperiostosis who demonstrated the whole syndrome (pachyderma, periostitis, and cutis verticis gyrata) is presented, and the Magnetic Resonance Imaging (MRI) appearances of the long bone and scalp changes are demonstrated. MRI of the cruris demonstrated fluffy periosteal new bone formation that encroached on the medullary cavity as well as expansion of the diaphysis. Cranial changes included thickening of the diploe associated with diminished signal of the intradiploic fat, and thickening of the scalp with furrowing.

Key words: Osteoarthropathy, primary hypertrophic, bones, scalp, Magnetic Resonance Imaging.

INTRODUCTION

Pachydermoperiostosis, also known as Touraine-Solente-Gole Syndrome, represents the primary (hereditary or idiopathic) form of hypertrophic osteoarthropathy. It constitutes only 3 to 5 per cent of all cases of hypertrophic osteoarthropathy. The clinical features of the syndrome include enlargement of the hands and feet, clubbing of the fingers, enlargement of the extremities secondary to soft tissue hypertrophy and periosteal new bone formation, coarsening of the facial features, and oily and thickened facial skin with furrowing [1, 2, 3]. We report the MRI imaging finding in a patient with this rare syndrome.

CASE REPORT

A 42 year-old-man was first admitted 8 years ago because of bone and joint pains. Fluffy, poorly defined periosteal new bone formation was detected in the long tubular bones of the upper and lower extremities.

On physical examination furrowing and thickening of the skin of the forehead, clubbing of the fingers and thickening of the skin of the hands and feet were noticed and the diagnosis of pachydermoperiostosis was made. In the family history, the patient claimed that his grandfather also had a furrowed forehead and ptosis of the eyelids. When he presented again 8 years later, thickening of the skin of the hands and feet were more prominent. Bone pains were more severe. Furrowing of the forehead had increased and extended to the scalp, On inspection, the scalp showed a gyral pattern (cutis verticis gyrata). He had ptosis and he could barely open his eyes. The periosteal reaction was also more prominent and exuberant bony excrescences were seen (figures 1a and 1b). An MRI examination was performed. MRI of the cruris demonstrated fluffy periosteal new bone formation that encroached on the medullary cavity, and expansion of the diaphysis (figure 2). MRI of the cranium demonstrated thickening of the diploe associated with diminished intradiploic fat signal, and thickening of the scalp with furrowing (figure 3).

DISCUSSION

The first cases of pachydermoperiostosis were reported by Friedreich in 1868 and they were consti-
red to be examples of acromegaly [4]. In 1935, Touraine, Solente and Gole defined the characteristics of this syndrome as a distinct entity and attracted attention to the similarities between the syndrome and pulmonary osteoartropathy [5]. It was Brugsch, who recognized the familial nature of the syndrome [6]. The syndrome has also been called idiopathic familial generalized osteophytosis, generalized hyperostosis with pachyderma, pachydermothyperostosis, acropachyderma with pachydermitis, osteodermatopathia hypertrophicans and Touraine-Solente-Gole Syndrome.

The syndrome is inherited with an autosomal dominant trait with variable expressivity, and this explains why all the patients do not demonstrate the whole syndrome (pachyderma, periostitis, cutis verticis gyrata). The scalp is spared in the incomplete form (cutis verticis gyrata is absent). The form fruste consists of pachyderma with minimal or absent periostitis. The syndrome has a predilection for men and the affected men demonstrate more severe symptoms than women. The symptoms begin in adolescence and progress slowly for about 10 years before the disease becomes self limited. The soft tissues of the hands and feet are enlarged, the distal ends of the fingers and toes are wide. The skin of the hands, feet and face is thickened, oily and, excessive sweating is seen. The facial features are coarse-

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<th>Fig. 1. — Conventional radiograms.</th>
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<td>A. Fluffy, ill-defined periosteal new bone formation along the medial cortex of the radius (arrows).</td>
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<tr>
<td>B. Endosteal new bone formation encroaching on the medullary cavity in the femur (arrows).</td>
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| Fig. 2. — Coronal spin-echo T1-weighted MR image. Periosteal (small arrows), and endosteal (large arrows) new bone formations are seen as hypointense signals. |

| Fig. 3. — Sagittal, spin-echo T1-weighted MR image. Thickening of the diploe associated with diminished signal of the intradiploic fat is demonstrated. Note furrowing of the scalp. |

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<td>A. Néoformation osseuse floconneuse à limites floues au niveau du cortical interne du radius (flèches).</td>
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<tr>
<td>B. La néoformation endostéale envahit la cavité médullaire du fémur (flèches).</td>
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| Fig. 2. — Coupe coronale spin-écho en T1. Néoformations osseuses périostéales (petites flèches) et endostéales visualisées en hyposignal. |

| Fig. 3. — Coupe sagittale spin-écho en T1. Épaississement de la diploé associé à une diminution du signal de la graisse intradiploïque. Notez la formation de sillon au niveau du cuir chevelu. |

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nerves or the spinal cord may result in neurologic abnormalities [1, 2, 3].

The clinical features of primary and secondary hypertrophic osteoarthropathy are similar. They are differentiated by family history and presence or absence of a primary lesion, such as a bronchogenic carcinoma. The radiographic findings also have significant differences. In pachydermoperiostosis, the periostial reaction is irregular and poorly defined bony outgrowths are seen in contradiction to the linear deposits seen with secondary hypertrophic osteoarthropathy. This more severe periostitis can be explained by the early onset and long duration of the disease [1, 7]. In our patient, the long bone changes, demonstrated by conventional radiography and MRI, included periosteal and endosteal new bone formation, and expansion of the diaphysis (figures 1 and 2).

The folded appearance of the skin of the scalp was named cutis verticis gyrata by Unna, in 1907 [8]. Grönberg was the first to recognize the association of Cutis verticis gyrata and periostitis [9]. Cutis verticis gyrata may be associated with systemic disorders such as acromegaly, myxedema, amyloidosis, pachydermoperiostosis or, it may be secondary to diseases of the scalp (tumors, neurofibromas, intradermal nevi, inflammatory conditions) or it may be a primary condition [10]. There are a few reports on CT and MRI of pachydermoperiostosis. CT demonstration of cutis verticis gyrata was first reported by dePadova [10], and Araki [11] demonstrated MR findings of pachydermoperiostosis in the skull and spine. MRI findings in our patient were thickening of the diploe associated with diminished intradiploic fat signal, and cutis verticis gyrata (figure 3).

REFERENCES