Case report

MR IMAGING OF PACHYDERMOPERIOSTOSIS

G. DEMIRPOLAT, R.N. SENER, E.E. STUN

Department of Radiology, Ege University Hospital, Bornova, Izmir, 35100-TR, Turkey.

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.

RéSUMÉ

Pachydermopériostose : imagerie par résonnance magnétique

Nous rapportons un cas de pachydermopériostose avec un tableau complet (pachydermie, périosite, cutis verticis gyrata) et présentons l’imagerie par résonnance magnétique (IRM) des modifications des os longs et du crâne. L’IRM des os longs a montré une néoformation osseuse floconneuse avec engoncissement du canal médullaire et une expansion au niveau diaphysaire. Au niveau crânien un épaississement de la diploé a été associé à une diminution de la graisse intradiploïque, ainsi qu’à un épaississement du cuir chevelu avec formation de sillons.

Mots-clés : ostéarthropathie, hypertrophie primaire, os, crâne, IRM.

INTRODUCTION

Pachydermoperiostosis, also known as Touraine-Solente-Gole Syndrome, represents the primary (hereditary or idiopathic) form of hypertrophic osteo-arthropathy. 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 MR 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 MR imaging 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 conside-
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, pachyder莫hyperostosis, acropachyderma with pachype-riostitis, 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-

Fig. 1. — Conventional radiograms.
A. Fluffy, ill-defined periosteal new bone formation along the medial cortex of the radius (arrows).
B. Endosteal new bone formation encroaching on the medullary cavity in the femur (arrows).

Fig. 1. — Radiogramme conventionnel.
A. Néoformation osseuse floconneuse à limites floues au niveau du cortical interne du radius (flèches).
B. La néoformation endostéale envahit la cavité médullaire du fémur (flèches).

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. 2. — Coupe coronale spin-écho en T1. Néoformations osseuses périostéales (petites flèches) et endostéales visualisées en hyposignal.

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.

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.

ned, and facial folds are more prominent. Joint pain or swelling is less common than with secondary hypertrophic ostearthropathy. Motion can be restricted secondary to perarticular osseous excrescences. Osseous compression on the cranial or peripheral
of the diploe associated with diminished intradiploic fat signal, and cutis verticis gyrata (figure 3).

REFERENCES


DIPLÔME UNIVERSITAIRE DE RADIOLOGIE
OTO-NEURO-OPTHALMOLOGIQUE

Organisé par les Professeurs
J.L. DIETEMANN, P. BOURJAT, F. VEILLON (Strasbourg)
L. PICARD, M. BRAUN, S. BRACARD (Nancy)
J.F. BONNETVILLE (BESANCON), D. BINNERT (DIJON)

L’enseignement comprend 6 semaines de cours et de travaux dirigés répartis sur deux années universitaires.

Les cours se déroulent les lundi et mardi pour les 2 premières sessions et les lundi, mardi et mercredi pour la 3e session à l’Institut de Radiologie de la Faculté de Médecine de Strasbourg (5e étage), 11, rue H umann, 67085 Strasbourg (Tél. 03 88 35 85 52). Les cours du matin débutent à 9 heures et ceux de l’après-midi à 14 heures 30.

Les cours se déroulent les lundi, jeudi et vendredi pour les 2 premières sessions et les jeudi et vendredi pour la 3e session à Nancy dans le service de N euroradiologie du Prof. L. Picard à H ôpital Neurol ogique — H ôpital Central — 29, avenue M aréchal de L attre de Tassigny 54035 NA N CY

Renseignements
Professeur J. L. DIETEMANN,
Service de Radiologie 2, H ôpitaux U niversitaires,
H ôpital de H autepierre
A venue M olière F-67098 Strasbourg Cedex
Tél. : (M me P hotais, Instutut de R adio) 03 88 35 85 52
Fax : 03 88 12 71 18
Professeur L. PICARD
Service de N euroradiologie
H ôpital neurol ogique — H ôpital Central
29, av. M aréchal de L attre de Tassigny 54035 NA N CY

© 2018 Elsevier Masson SAS. Tous droits réservés. - Document téléchargé le 30/10/2018 Il est interdit et illégal de diffuser ce document.