Case 14477

Eurorad

McCune Albright Syndrome (MAS) – polyostotic fibrous dysplasia

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Section: Musculoskeletal system
Area of Interest: Musculoskeletal bone Musculoskeletal joint Musculoskeletal system
Procedure: Education
Imaging Technique: Conventional radiography
Imaging Technique: MR
Imaging Technique: SPECT
Special Focus: Congenital Metabolic disorders Case Type: Clinical Cases
Authors: Lukasz Augsburg1, Filip M. Vanhoenacker 1, 2, 3, Jan Gielen1
Patient: 15 years, male

Clinical History:

A 15-year-old boy presented to the paediatrician with precocious puberty, hyperthyroidism, hyperprolactinaemia and large, asymmetric head.
Inspection of the skin showed “café-au-lait” spots located mainly on the back (Fig. 1) and left buttock.
Imaging Findings:

Conventional radiography (CR) showed multiple bone lesions with a ground glass appearance in both femurs (Fig. 2, 3), left humerus (Fig. 4), right tibia and right talus (Fig. 5, 6). Bone scintigraphy showed multifocal tracer uptake in the axial and appendicular skeleton suggesting active osteoblastic activity (Fig. 7).
Magnetic Resonance Imaging (MRI) of the brain showed asymmetric expansion of the calvaria and facial bones with significant bone thickening. Lesions showed predominantly low, slightly inhomogeneous signal intensity on all pulse sequences (Fig. 8, 9, 10). The brain was normal (Fig. 8, 9). The orbits were small. (Fig. 10). However, there was no compression of the optic nerve (Fig. 10).
Discussion:

Based on the combination of clinical and imaging findings, the diagnosis of McCune-Albright syndrome (MAS) was made.
MAS is a genetic disorder characterized by a typical triad of symptoms: polyostotic fibrous dysplasia (FD), skin pigmentation (café-au-lait spots) and endocrinological disorders [1]. Gonadal dysfunction is considered to be the most common endocrine defect in females, resulting in precocious puberty and vaginal bleeding. However, Cushing syndrome, pituitary adenomas and hyperthyroidism are also reported [2]. MAS develops from activation of a post-zygotic, sporadic or somatic mutation of the GNAS1 gene with different clinical manifestations [3]. Affected children usually present with skeletal deformities and short stature, resulting from premature closure of the epiphyses.
FD is a tumour-like condition of the bone. Histologically it corresponds to immature bone with delicate trabeculae and interspersed fibrous matrix [4]. This makes the bone more prone to microfractures, repetitive remodelling and subsequent deformation [5]. FD associated with MAS usually affects multiple bones and leads more frequently to complications such as fractures, skeletal deformities or limb-length discrepancies. It affects most commonly the

femora, tibiae, pelvis, skull and ribs. Imaging features of FD may vary from either completely radiolucent lesions, sclerotic delineated radiolucent lesions or lesions with a ground-glass matrix appearance. In case of disease progression, bowing of the proximal femora (coxa vara) may occur due to multiple fractures and repetitive remodelling, resulting in the so-called shepherd's crook deformity. [4, 6].
Conventional radiography (CR) is usually sufficient to make a reliable diagnosis.
Bone scintigraphy may reveal increased osteoblastic activity in affected bones. It is particularly useful to assess multifocality.
Computed tomography (CT) is usually not mandatory, but may be used to evaluate the precise extent of FD and potential compression of adjacent structures in complex anatomical structures such as the facial bones, skull base and the axial skeleton. However, due to high radiation dose, the use of CT should be considered with caution.
Particularly in the paediatric population, MRI is the preferred technique for evaluation of the lesion extent in the skull bones. Bone expansion may cause compression of adjacent structures, such as the optic nerve, resulting in visual disturbances. Osteotomy of the involved bones should be considered in this scenario.
Malignant transformation of bone lesions is rare.
Paget's disease may appear similar on imaging but is seen in older patients. Association of FD and intramuscular myxoma is known as Mazabraud syndrome.
Differential Diagnosis List: Polyostotic fibrous dysplasia related to McCune-Albright Syndrome, Mazabraud syndrome, Sporadic fibrous dysplasia, Paget's disease

Final Diagnosis: Polyostotic fibrous dysplasia related to McCune-Albright Syndrome

References:

Rao S, Colaco MP, Desai MP (2003) McCune Albright Syndrome (MCAS): a case series. Indian Pediatr 40(1):29–35. (PMID: 12554915)
Rubio NI, Nader Sh, Brosnan PG (2006) McCune Albright syndrome: case report and review of literature. Int J Endocrinol Metab 4(3):167–75
Diaz A, Danon M, Crawford J (2007) McCune-Albright syndrome and disorders due to activating mutations of GNAS. J Pediatr Endocrinol Metab 20(8):853–80 (PMID: 17937059)
Xavier SP, Ribeiro MC, Sicchieri LG, Brentegani LG, Lacerda SA (2008) Clinical, microscopic, and imaging findings associated to McCune-Albright syndrome: A report of two cases. Braz Dent J 19: 165 – 170. (PMID:18568233)
Martin-Carreras T, Sanchez E, Bancroft LW (2014) Polyostotic fibrous dysplasia. Orthopedics Nov 1. 37(11):722-82 (PMID: 25361354)
Medow JE, Agrawal BM, Resnick DK (2007) Polyostotic fibrous dysplasia of the cervical spine: case report and review of the literature. Spine J 7: 712 – 715 (PMID: 17434805)

Figure 1

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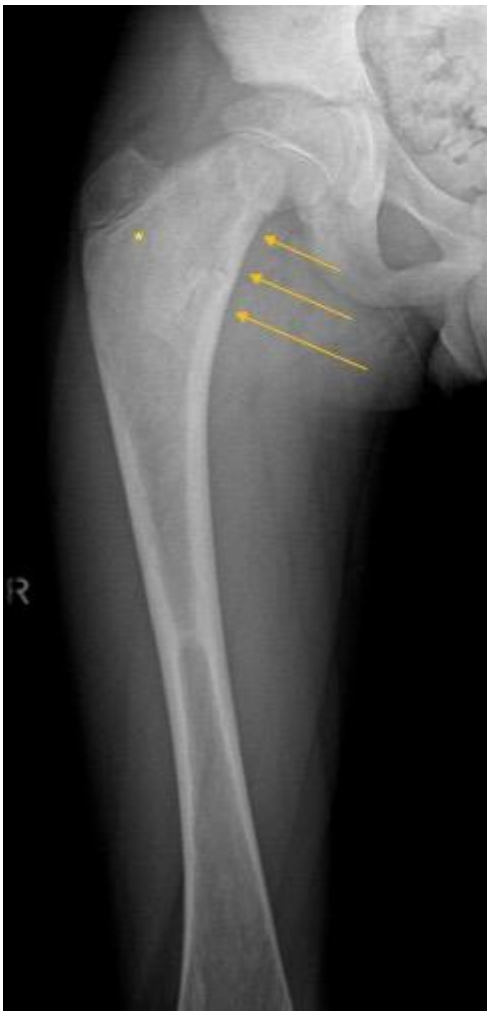

Description: Conventional radiography of the right femur (AP projection) shows a slightly expansile lesion (asterisk) in the proximal diaphysis with a ground-glass appearance and widening of the medullary cavity. Note subtle deformity of the hip (arrows). **Origin:** Augsburg L, Department of Radiology, UZA, Antwerp, Belgium

Figure 2

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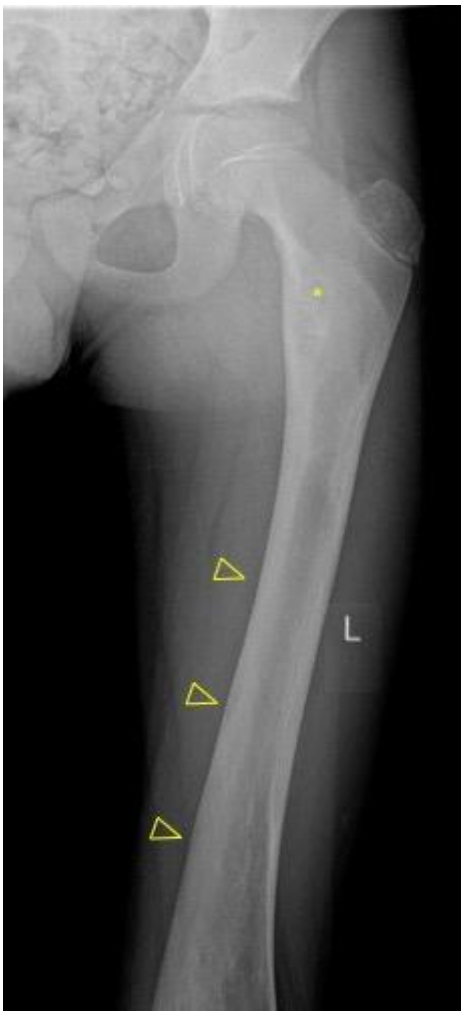

Description: Conventional radiography of the left femur (AP projection) shows a ground-glass lesion in the proximal diaphysis. Note also more subtle lesions in the middle and distal diaphysis (arrowheads). Note slight deformity of the femoral diaphysis. **Origin:** Augsburg L, Department of Radiology, UZA, Antwerp, Belgium

Figure 3

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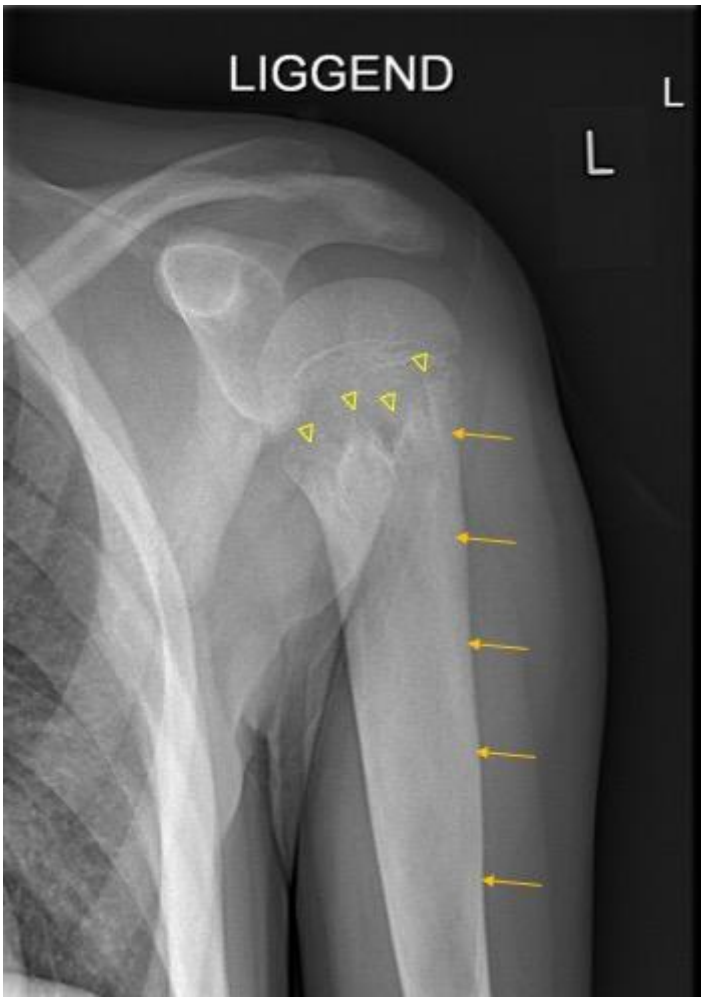

Description: Conventional radiography of the left humerus (AP projection) shows bone expansion of the proximal diaphysis of the left humerus with homogeneous ground-glass matrix (arrows). Note sclerotic teeth-like border with normal proximal metaphysis (arrowheads). **Origin:** Augsburg L, Department of Radiology, UZA, Antwerp, Belgium

Figure 4

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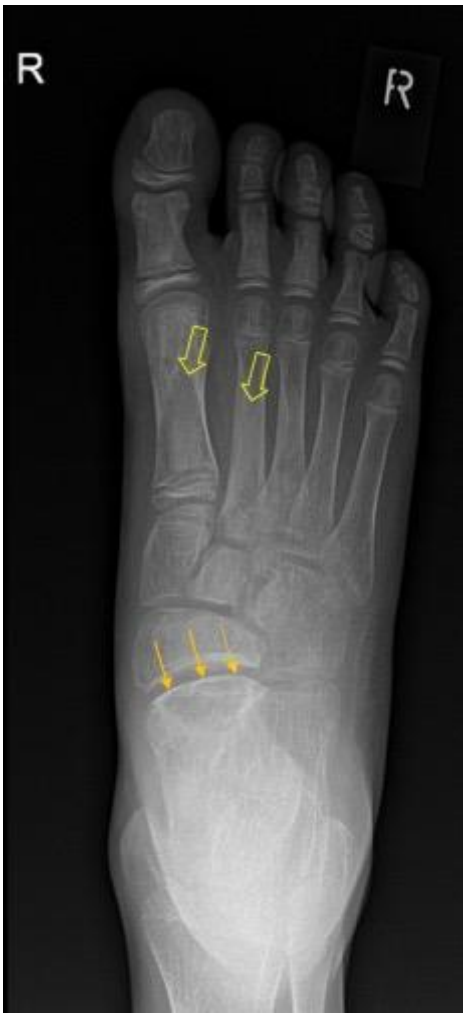

Description: Conventional radiography of the right foot (AP projection) depicts a focal radiolucent lesion with sclerotic margin in the talar head (arrows). Note also lesions with ground-glass aspect in the metatarsals (thick open arrows). **Origin:** Augsburg L, Department of Radiology, UZA, Antwerp, Belgium

Figure 5

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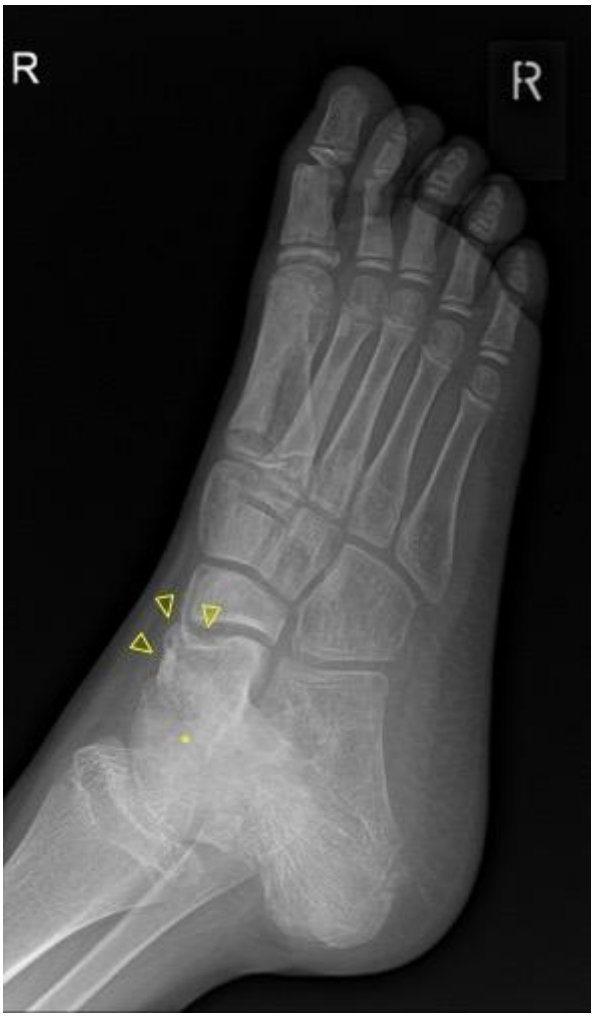

Description: Conventional radiography of the right foot (oblique projection) depicts the ground-glass appearance in the talar body and neck (asterisk) and deformity of the head (arrowheads).**Origin:** Augsburg L, Department of Radiology, UZA, Antwerp, Belgium

Figure 6

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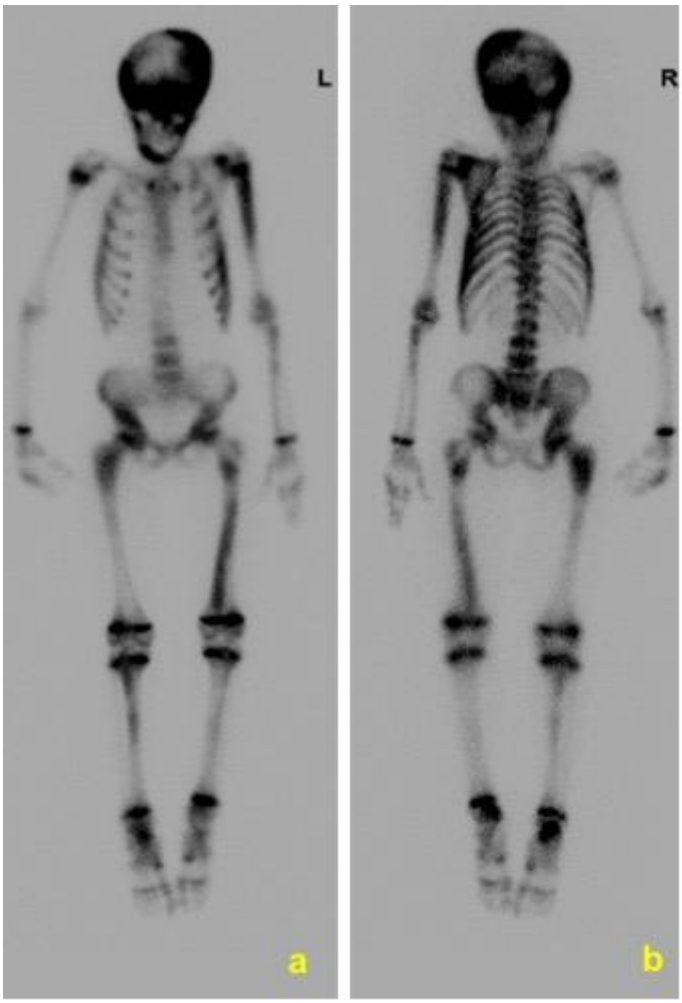

Description: Bone scintigraphy. Anterior (a) and posterior (b) projection show increased osteoblastic activity in the skull, ribs, pelvis, left humerus, both femora, right tibia and talus. **Origin:** Augsburg L, Department of Radiology, UZA, Antwerp, Belgium

Figure 7

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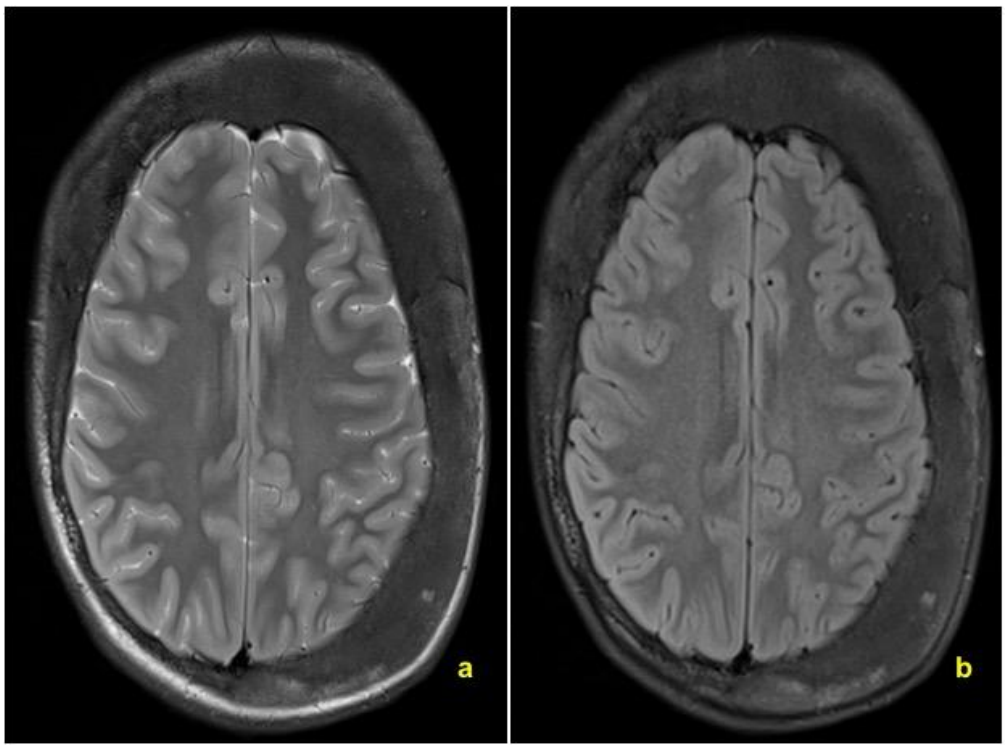

Description: MR of the brain. Axial T2-weighted images (a), FLAIR images (b). MR shows marked but asymmetrical thickening of the calvaria. Although the lesions are slightly inhomogeneous, the overall appearance is of predominant low signal. **Origin:** Augsburg L, Department of Radiology, UZA, Antwerp, Belgium

Figure 8

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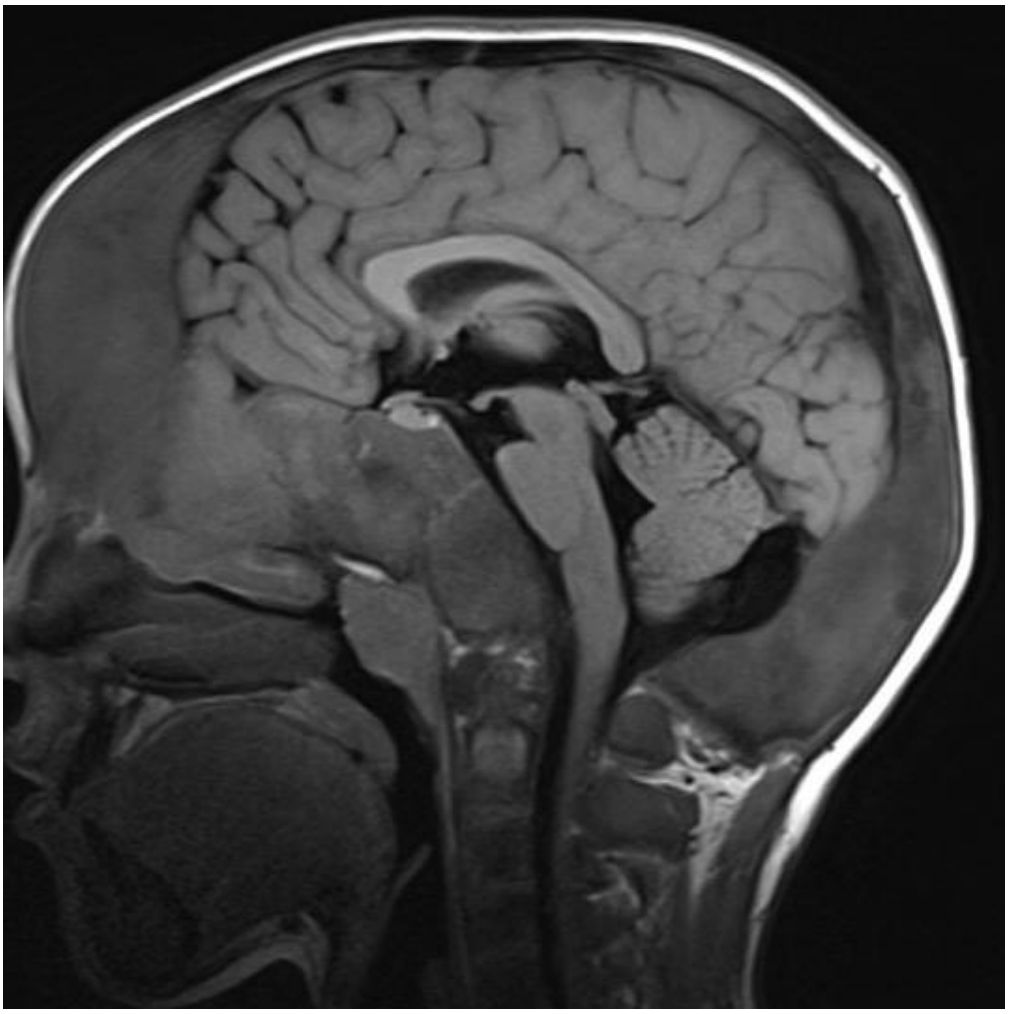

Description: MR of the brain. The mid-sagittal T2- weighted image demonstrate marked thickening of frontal-, occipital-, sphenoid bone and clivus. Intracranial structures are normal. **Origin:** Augsburg L, Department of Radiology, UZA, Antwerp, Belgium

Figure 9

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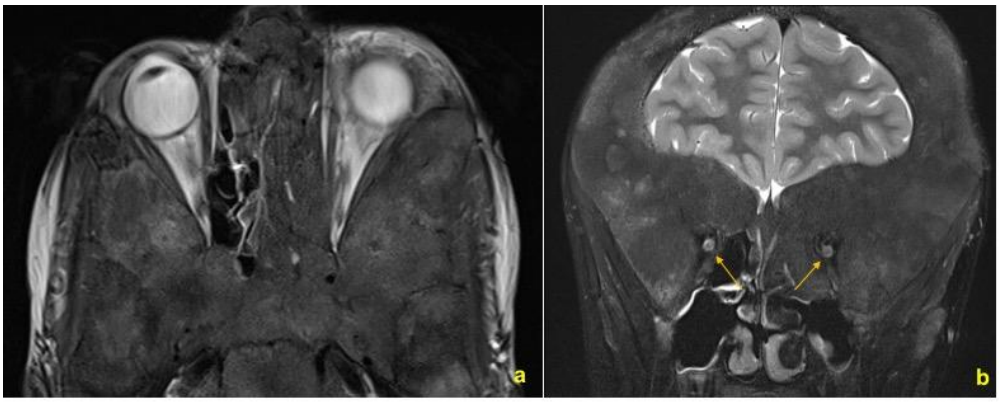

Description: MR of the brain. The axial T2 weighted image (a) show narrowing of the orbit. There is no definite compression of the optic nerve (arrow) on the coronal image (b). **Origin:** Augsburg L, Department of Radiology, UZA, Antwerp, Belgium

Figure 10

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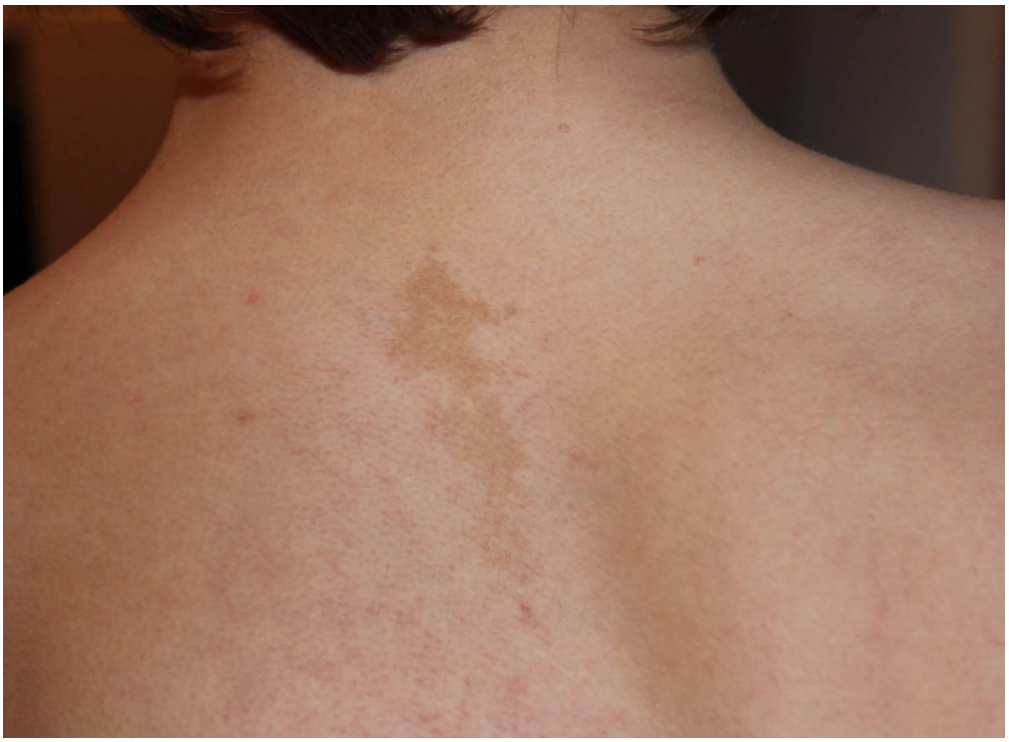

Description: Irregularly delineated café-au-lait spots, resembling the coast of Maine in the United States of America. **Origin:** Lukasz Augsburg, UZA, Antwerpen