MCCUNE-ALBRIGHT-STERNBERG

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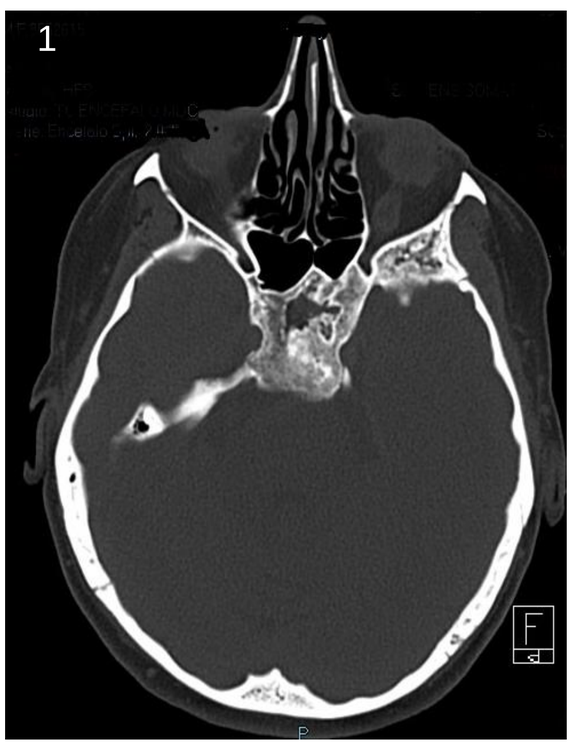
INTRODUCTION

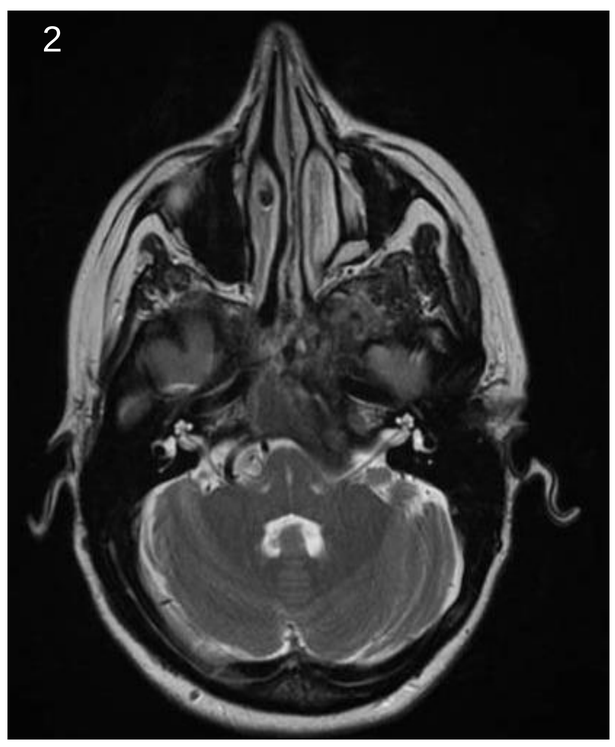
Fibrous dysplasia/McCune-Albright syndrome (FD/MAS), the result of an embryonic postzygotic somatic activating mutation of *GNAS* (encoding the cAMP pathway-associated G-protein, $G_s\alpha$), is characterized by involvement of the skin, skeleton, and certain endocrine organs. However, because $G_s\alpha$ signaling is ubiquitous additional tissues may be affected. MAS classically defined by the clinical triad of fibrous dysplasia of bone (FD), café-au-lait skin spots, precocious puberty (PP). It is a rare disease with estimated prevalence between 1/100,000 and 1/1,000,000

CASE REPORT

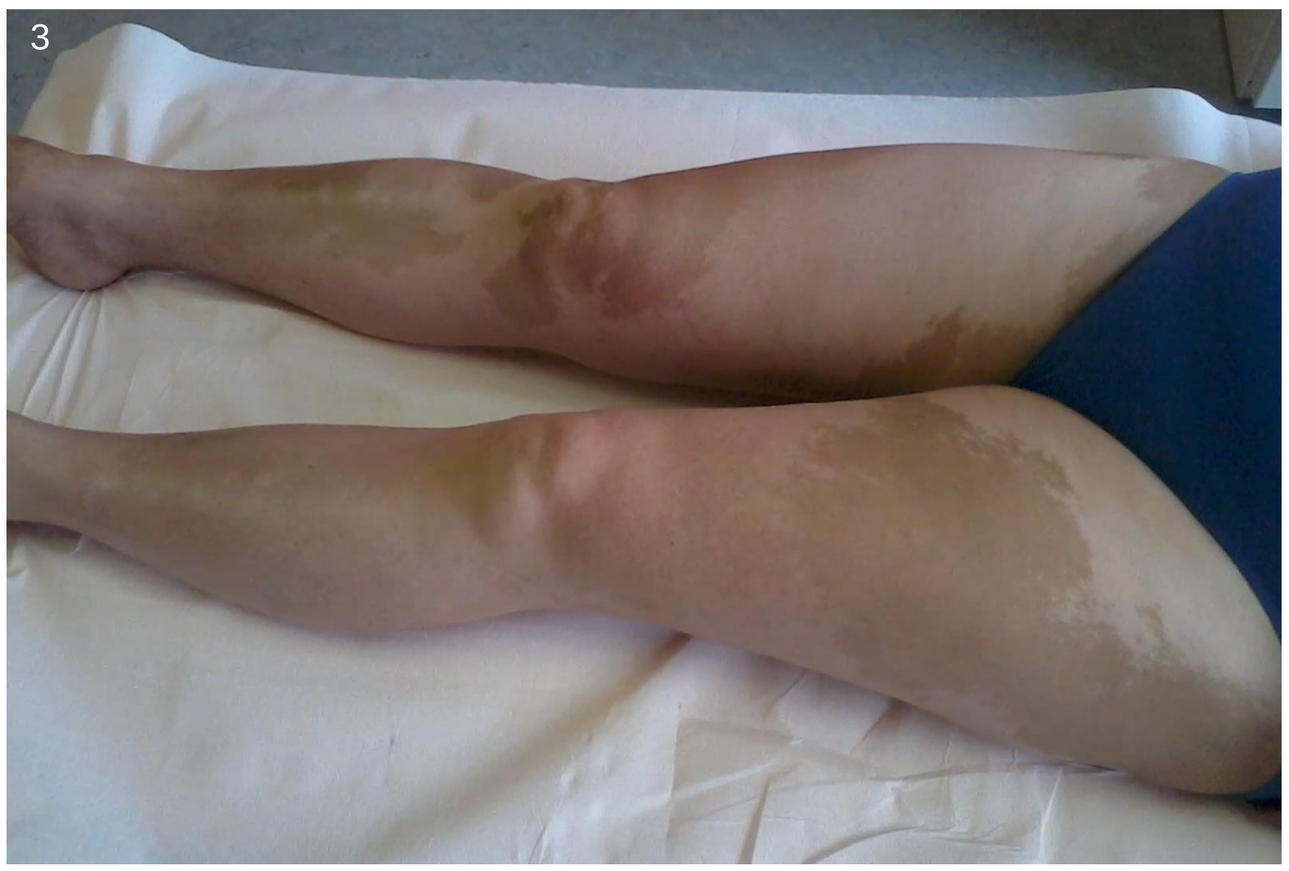
We report the case of a 48 years old caucasian woman evaluated due to a previous diagnosis of persistent migraine without aura. She complained of occipital oppressive pain spreading to the whole scalp associated to photophobia from many years, partially responsive to common non steroideal anti-inflammatory drugs (NSAIDS).
Her medical history was positive for congenital café-au-lait spots from the abdomen involving legs bilaterally and diagnosis of pituitary dwarfism diagnosed when she was 10, treated with growth hormone, hypercholesterolemia. Puberty occurred when she was 11.
Her neurological examination was normal; general examination showed short stature (149 cm) bilateral winged scapula, scoliosis, varus deformity, flat-feet, bilateral xanthelasma around the eyelids.

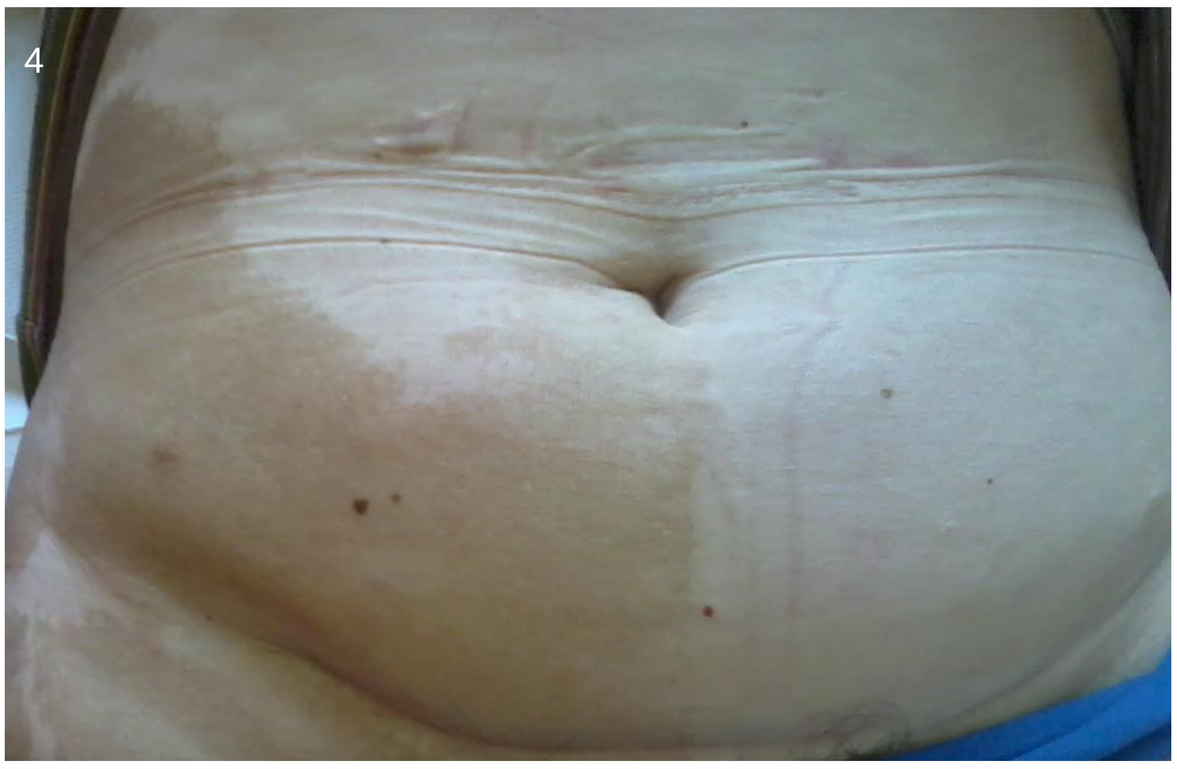
Figure 1: brain CT showing fibrous dysplasia
Figure 2: brain MRI schowing fibrous dysplasia





Figures 3-4: cafè au lait skin spots involving trunk and legs





RESULTS

She performed brain CT and MRI showing hypointense areas on T1-weighted images involving expecially occipital bone clivus, left sphenoid bone body (cotton wool appearance), pterygoid process, great wing of the left sphenoid (pterygopalatine fossa), consistent with radiographic presentations of fibrous dysplasia. Skeletal radiography didn't show other areas of fibrous dysplasia. Hormone testing andurinary chatecolamines showed mild vitamine D (25 OH) insufficiency.
She performed skin biopsy (café-au-lait leg) and peripheral blood genetic analysis (selective amplification of the mutant allele) which identified the presence of the mutation **c.604C> T,** with a RMA (Relative Abundance Mutation vs internal reference lab) in the sample tested about 3-6% in the biopsy and about 25% in the peripheral blood. This result is compatible with the presence of the mutation associated with MAS, even if the method has yet to be validated.

CONCLUSION

FD/MAS is a rare, genetic disorder which can be painful, expecially due to to obstructed paranasal sinuses or compression of nerves in foramina. The management of craniofacial FDs is often medical (NSAIDS) but may become strictly surgical in case of cosmetic deformation, intractable pain, disease progression. We think that cranial fibrous dysplasia has to take into account as a possible differential diagnosis when evaluating a persistent head pain in young people only temporarily responsive to NSAIDS.

BIBLIOGRAPHY

- McCune-Albright syndrome and the extraskeletal manifestations of fibrous dysplasia. Collins MT, Singer FR, Eugster E. Orphanet J Rare Dis. 2012 May 24;7 Suppl 1:S4.
- McCune-Albright syndrome in adulthood Chanson P, Salenave S, Orcel P. Pediatr Endocrinol Rev. 2007 Aug;4 Suppl 4:453-62.
- McCune-Albright Syndrome: An Overview of Clinical Features. Brillante B, Guthrie L, Van Ryzin C. J Pediatr Nurs. 2015 Sep-Oct;30(5):815-7