

CASE REPORTS

The “Phantom of the Opera” sign of craniofacial fibrous dysplasia with unilateral polyostotic involvement in McCune-Albright syndrome

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ABSTRACT

McCune-Albright syndrome is a very rare genetic disorder resulting from a sporadically occurring somatic *GNAS* gene mutation. It is characterised by the association of: endocrinopathy (most commonly precocious puberty), polyostotic fibrous dysplasia, and cutaneous pigmentation with café-au-lait spots with edges resembling the coast of Maine. We present a case of McCune-Albright syndrome with unilateral polyostotic fibrous dysplasia including involvement of the skull and facial bones on one side resulting in an appearance on skeletal scintigraphy resembling the characteristic mask of the “Phantom of the Opera”.

Key Words: Fibrous dysplasia, Craniofacial, McCune-Albright, Bone scan

1. INTRODUCTION

Fibrous dysplasia is a common benign bone disorder with monostotic and polyostotic forms. However, McCune-Albright syndrome – of which polyostotic fibrous dysplasia is a component – is a very rare condition. The fibrous dysplasia in McCune – Albright syndrome is generally more severe than in sporadic cases and typically involves or predominates on only one side of the body.

2. CASE REPORT

We report the case of a 49-year-old Caucasian male with McCune-Albright syndrome diagnosed during childhood, who presented for investigation of back pain. Technetium-^{99m} (^{99m}Tc) hydroxymethoxydiphosphonate (HDP) bone scan showed intense abnormal radiotracer uptake in multiple bones on the left side of the body (see Figures 1 and 2) involving the skull and facial bones, left second and eighth ribs,

which were also present and unchanged on a bone scan from four years prior. Computed tomography (CT) and single-photon emission tomography (SPECT) were also performed at the time of the bone scan demonstrating marked expansion of the left frontal skull and maxilla with ground glass internal matrix characteristically seen in fibrous dysplasia (see Figure 3). There was a small amount of contiguous extension into the right frontal bone, with the overall distribution of craniofacial disease resembling the mask worn by the “Phantom of the Opera” (see Figure 1 inset), the villain in a novel by Gaston Leroux more recently popularised by its musical adaptation by Andrew Lloyd Webber. There was also a new finding of linear increased radiotracer uptake in the superior endplate of the T12 vertebral body that correlated with a low-grade compression fracture on CT and likely accounted for the patient’s back pain.

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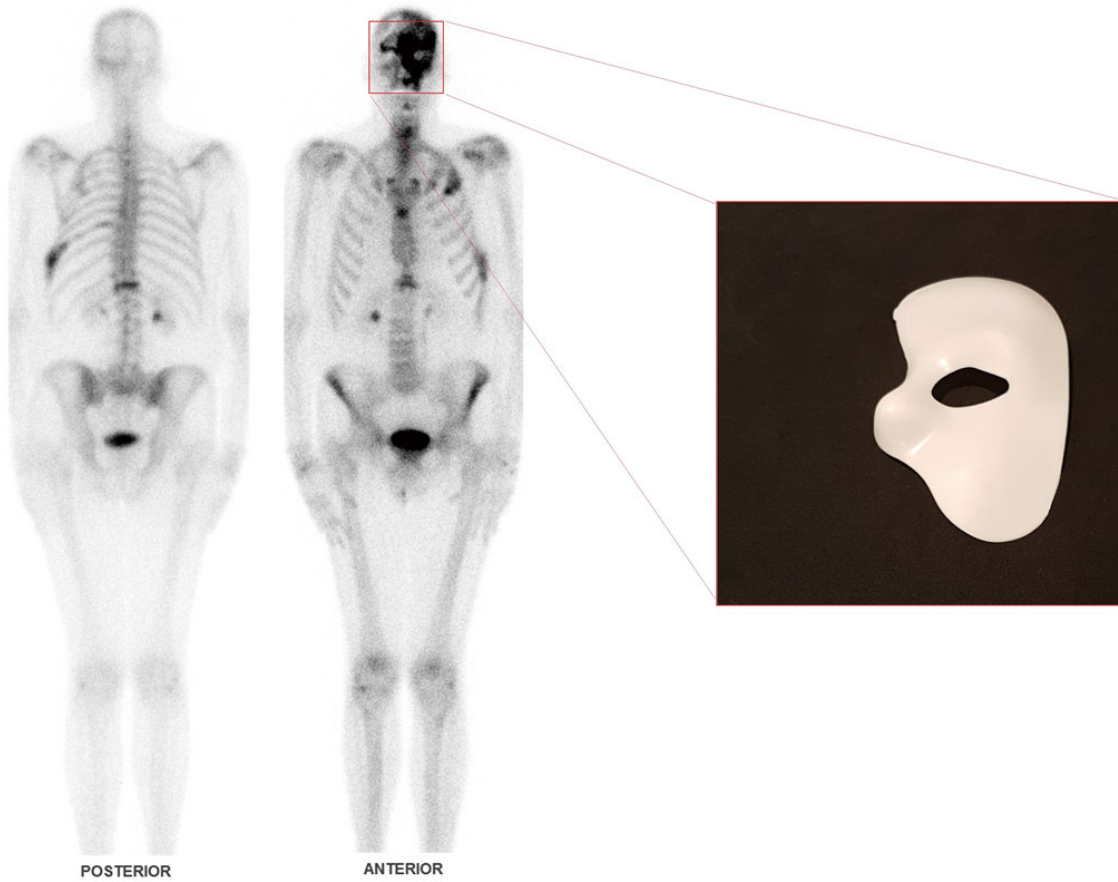


Figure 1. ^{99m}Tc -HDP bone scan whole body anterior and posterior planar views; inset, mask of the “Phantom of the Opera”

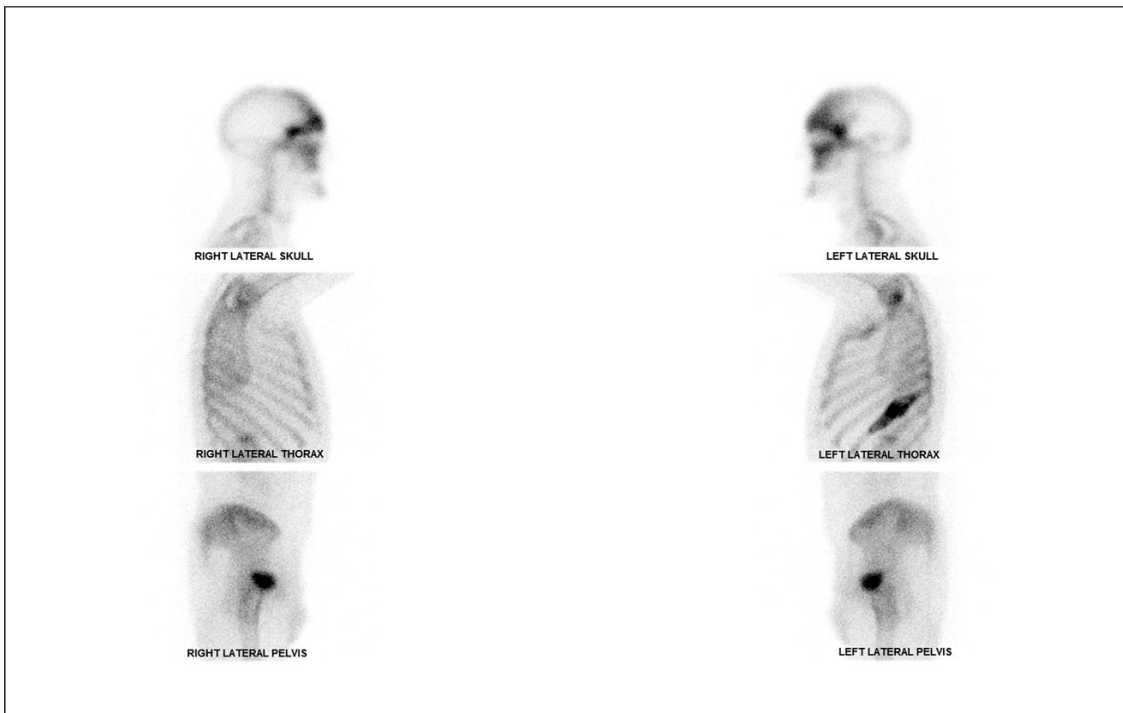


Figure 2. ^{99m}Tc -HDP bone scan lateral static views of the skull, thorax and pelvis

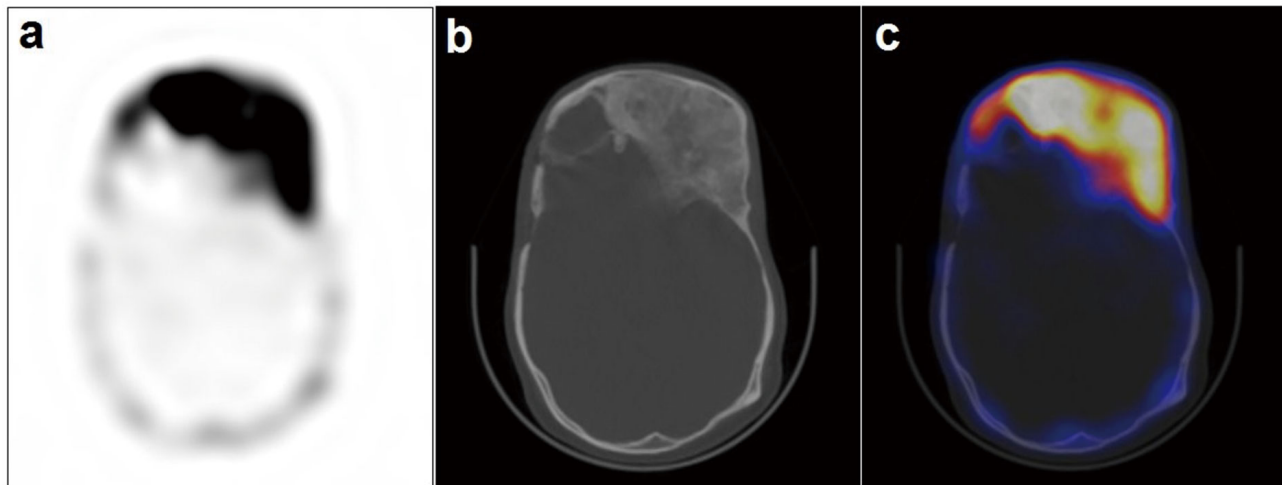


Figure 3. a. SPECT, b. CT, and c. Fused SPECT/CT showing marked expansion of the left frontal skull and maxilla with ground glass internal matrix characteristically seen in fibrous dysplasia

3. DISCUSSION

McCune-Albright syndrome results from an activation mutation of the *GNAS1* gene that causes overproduction of a variety of protein products.^[1] It is a very rare condition. All reported cases are sporadic with no known hereditary basis. The syndrome is characterised by at least two of the following three features: (1) polyostotic fibrous dysplasia, (2) café-au-lait pigmented skin lesions, (3) autonomous endocrine hyperfunction.

Polyostotic fibrous dysplasia. Fibrous dysplasia refers to bone that is replaced by abnormal fibrous connective tissue, with the affected bones weakened, fragile and prone to pathological fracturing. In McCune-Albright syndrome, fibrous dysplasia is invariably polyostotic, generally more severe than in sporadic cases, and involves or predominates on only one side of the body.^[2]

Pigmented skin lesions. The dermal lesions are flat macules that typically have a brown pigmentation (hence the term “café-au-lait” spots) and jagged borders that resemble the coast of Maine in north-eastern United States, frequently occurring on the back of the trunk (in a third to half of cases). There is a predominance of skin and bone lesions on the same and only one side of the body.

Endocrinopathy. The autonomous endocrine hyperfunction in McCune-Albright syndrome most commonly takes the form of precocious puberty that is gonadotropin-independent. Acromegaly is observed in 20%-30% of patients (as in the present case), and hyperthyroidism and Cushing syndrome

have also been described.^[3]

4. CONCLUSION

Fibrous dysplasia is a major component of McCune-Albright syndrome and differs from sporadic (non-syndromic) cases of fibrous dysplasia in its:

- severity (larger and more extensive in McCune-Albright syndrome), and
- distribution (typically predominating on only one side of the body in McCune-Albright syndrome).

The appearance of multiple osteoblastically active skeletal lesions on bone scan carries a differential diagnosis of metastases, polyostotic Paget’s disease and fractures (if there is a relevant clinical history), however, the unilaterality of osseous involvement should alert the clinician to the possibility of a syndromic cause for fibrous dysplasia, particularly if the other features of McCune-Albright syndrome are also present, namely:

- café-au-lait skin pigmentation affecting the same side of body as the osseous lesions, and
- autonomous endocrine hyperfunction – most commonly precocious puberty – with acromegaly also reported in 20%-30% of patients with the syndrome.

CONFLICTS OF INTEREST DISCLOSURE

The authors declare that they have no conflict of interest related to this manuscript.

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