

## It's not McCune-Albright Syndrome, It's Neurofibromatosis-1

Abdalla EM<sup>1\*</sup>, Nabil KM<sup>2</sup> and El-Hady GM<sup>1</sup>

<sup>1</sup>Department of Human Genetics, Medical Research Institute, Alexandria University, Egypt

<sup>2</sup>Department of Ophthalmology, Faculty of Medicine, Alexandria University, Egypt

A 9-year-old girl presented with severe progressive scoliosis. She was born to healthy Egyptian non-consanguineous parents and had normal growth and development. Physical examination showed cutaneous manifestations in the form of multiple oval, well circumscribed macular café-au-lait spots of varying sizes (A and D) and extensive disfiguring dark-brown patches with irregular borders, which were raised and hairy in some sites (B and C). These skin lesions were present at birth but they increased in size and number during early childhood. Scoliosis was the main health problem in the patient (D). A corrective surgery was performed at age of 6 but the problem continued its progression (operation scar is shown on panel D). Slit lamp examination detected Lisch iris nodules (E).

Neurofibromatosis-1 (NF 1) is a relatively common autosomal dominant disorder caused by a mutation in the NF1 gene encoding neurofibromin. A diagnosis of NF1 is made when two or more of the seven diagnostic features are present. These criteria include six or more café-au-lait macules (CALMs), axillary or inguinal freckles, two or more typical neurofibromas or one plexiform neurofibroma, optic nerve glioma, two or more Lisch nodules, distinctive osseous lesion such as sphenoid wing dysplasia, and having a first-degree relative with NF1. The presence of multiple CALMs is the first feature noticed in children and other features do not appear until later, delaying the diagnosis. Cutaneous neurofibromas, which are found in the majority of NF1 individuals, are rare in early childhood. Scoliosis may become evident in childhood or adolescence and girls are affected rather more often than boys. If it starts before the age of 10 years, scoliosis has a poor prognosis and is likely to be rapidly progressive.

McCune-Albright syndrome (MAS) is a sporadic disease caused by post-zygotic mutations in the GNAS1 gene. Classically, MAS comprises a triad of CALMs, fibrous dysplasia of bone and endocrine abnormalities such as precocious puberty. In MAS, the hyperpigmented skin lesions

are often few but can be quite large. They display a segmental distribution and demonstrate jagged borders with a tendency to predominate on one side of the body and to respect the midline.

Generally, CALMs in NF tend to be smaller and have a smooth outline ("coast of California"); whereas in MAS they are characterized by an irregular outline ("coast of Maine"). However, this distinction may not hold true in all cases. In this report, although the patient showed giant CALMs with irregular jagged borders and cutaneous neurofibromas were not present, ophthalmic assessment confirmed the diagnosis of NF1. The coexistence of multiple CALMs and Lisch iris nodules, constituted the basis of this diagnosis.

In conclusion, the presented case highlights the importance of a comprehensive clinical examination in establishing a correct diagnosis, which in this case could have been missed for several years. Slit-lamp examination may provide essential diagnostic information in older children and adults who present with only one clinical criterion such as multiple CALMs.

### Financial Disclosure

The authors indicate they have no financial relationships relevant to this article to disclose.

### Competing Interest

The authors declare that they have no competing interests.



Figure 1:

\*Corresponding author: Abdalla EM, Department of Human Genetics, Medical Research Institute, Alexandria University, 165 El-Horreya Avenue El-Hadra, Postal code 21561, Alexandria, Egypt, Tel: (+203) 4285455 - 4282373- 4288233; Fax: (+203) 4283719; E-mail: [ebtesam.nasr@alex-mri.edu.eg](mailto:ebtesam.nasr@alex-mri.edu.eg)

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