Kyphoscoliosis: looking beyond the spine

Joana Fonseca Ferreira, Margarida Coutinho, Cátia Duarte, José da Silva

Rheumatology Unit, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal

Correspondence to
Dr Joana Fonseca Ferreira, joana.fonseca.ferreira@gmail.com

Accepted 13 February 2015

DESCRIPTION

A 38-year-old woman was referred to the rheumatology department due to mechanical low back pain and progressive scoliosis that started in adolescence. She had no other joint or systemic symptoms despite pigmented skin lesions scattered throughout the body.

The physical examination revealed severe dorsal kyphosis associated with structural lumbar scoliosis and numerous 'café-au-lait' lesions (figure 1) spread over the skin, freckling in the arm pits and inguinal folds (figure 2). The radiographs revealed spinal misalignment, without fractures or lytic lesions. No relevant changes were observed in laboratory tests (figure 3).

A diagnosis of neurofibromatosis type 1 (NF1) was established.[1] Genetic testing revealed a mutation on NF1 gene, confirming the diagnosis.

One year later, the patient presented with persistent frontal headache, resistant to acetaminophen and non-steroidal anti-inflammatory drugs. A cranial CT scan revealed bilateral thickened optic nerves, suggesting optic gliomas (figure 4), a hallmark of this disease.

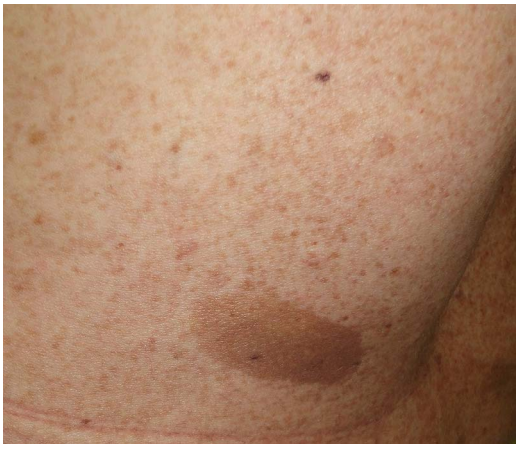

Figure 1 Dorsal 'cafe-au-lait' skin lesion.

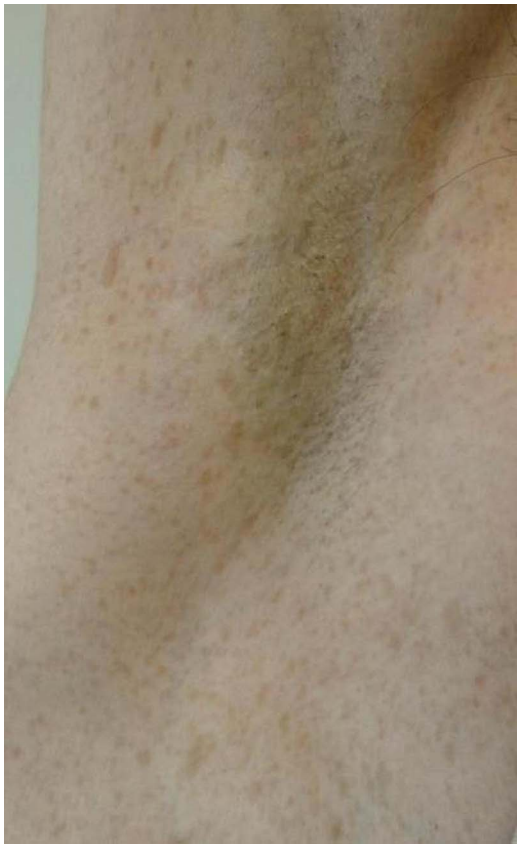

Figure 2 Armpit lesions: axillary freckling.

To cite: Ferreira JF, Coutinho M, Duarte C, *et al*. *BMJ Case Rep* Published online: [*please include* Day Month Year] doi:10.1136/bcr-2014-209096

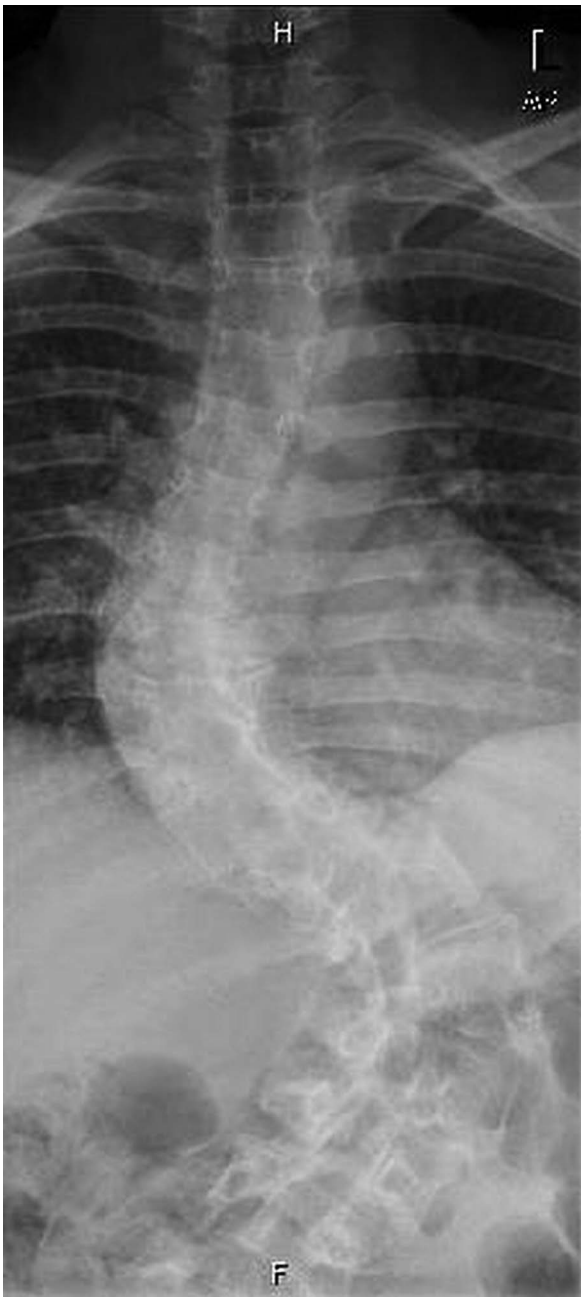

Figure 3 Anteroposterior X-ray exhibiting an exuberant dorsolumbar scoliosis.

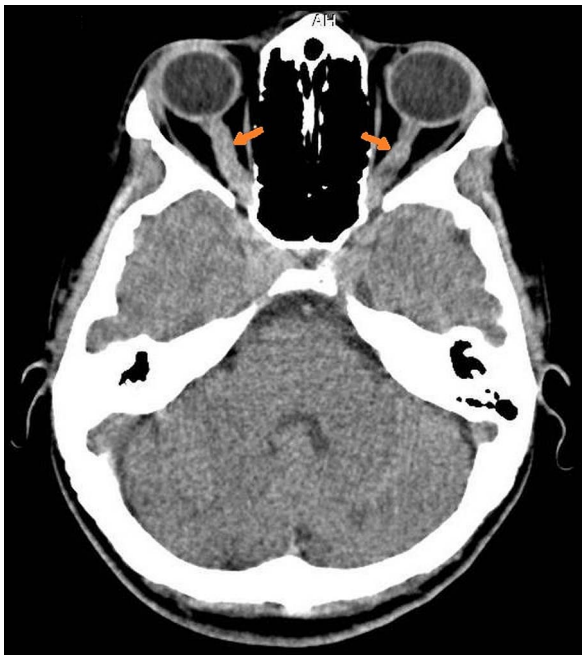

Figure 4 Cranial CT scan revealing bilateral thickening of the optic nerves (arrows), suggesting the presence of optic gliomas.

Learning points

- Neurofibromatosis type 1 (NF1) is an autosomal dominant disease, mainly characterised by the presence of 'café-au-lait' spots and neurofibromas (dermal being the most common).[2]
- NF1 can have cardiovascular, ophthalmological and osteoarticular manifestations, including kyphoscoliosis (occurring in approximately 10% of the cases), short stature and sphenoid wing dysplasia. The association of typical skin manifestations should suggest the diagnosis and prompt appropriate investigation.[3]
- There is no effective medical treatment to prevent or reverse the characteristic lesions of NF1, but an early diagnosis allows genetic counselling and early detection of treatable complications.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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