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CASE REPORT

Intracranial arterial dolichoectasia and skull damage in a girl with Jaffe-Campanacci syndrome: a case report

Yong Han[1] · Hangzhou Wang[1]

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Abstract

Jaffe-Campanacci is a rare syndrome characterised by axillary freckles, multiple non-ossifying fibromas of the long bones and jaw, and café-au-lait spots, associated with some features of neurofibromatosis type 1 (NF1). Cherix et al. and Colby and Saul suggested that Jaffe-Campanacci syndrome (JCS) might be a distinct form of NF1. Intracranial arterial dolichoectasia (IADE) is defined as an increase in the length and diameter of at least one intracranial artery. Affected intracranial arteries are dilated, elongated and sometimes tortuous. But in this rare disease of JCS, neither skull damage nor IADE has been previously reported. Here, we introduce the case of an 11-year-old Chinese girl with IADE, skull damage and features of JCS.

Keywords Jaffe-Campanacci syndrome · Intracranial arterial dolichoectasia · Skull damage

Dear Editor:

Jaffe-Campanacci is a rare syndrome characterised by axillary freckles, multiple non-ossifying fibromas of the long bones and jaw, café-au-lait spots, associated with some features of neurofibromatosis type1 (NF1) [1, 2]. Cherix and Colby et al. suggested that Jaffe-Campanacci syndrome (JCS) might be a distinct form of NF1 [3–5]. Intracranial arterial dolichoectasia (IADE) is defined as an increase in the length and diameter of at least one intracranial artery. Affected intracranial arteries are dilated, elongated and sometimes tortuous [6, 7]. But in this rare disease of JCS, neither skull damage nor IADE has been previously reported. Here, we introduce the case of an 11-year-old Chinese girl with IADE, skull damage and features of JCS.

An 11-year-old girl came to our hospital because of intermittent nausea and vomiting for half a year. CT examination of the head revealed occipital bone destruction which shows wormlike changes and sporadic calcification in the brain tissue (Fig. 1). Then a whole brain digital subtraction angiography (DSA) was performed. The result of DSA revealed IADE in this patient (Fig. 2). Examination of the skin revealed café-au-lait spots on her trunk and face (Fig. 3). Bilateral axillary freckles were also found, while neurofibroma was not observed. X-ray examination of the child's limbs revealed multiple bone lesions including right distal femur, both proximal tibias, right proximal fibula and left distal femur. These lesions were rounded, of variable size, well defined, mostly with sclerotic margins, corresponding to typical multiple nonossifying fibromas (Fig. 4). These bone lesions resulted in the inconsistent length of her legs. Then the surgery by open reduction was performed to treat these lesions. The diagnosis of multiple nonossifying fibromas was confirmed by pathological examination after surgery (Fig. 4). The bone lesions healed without complications after surgery, and the patient returned to school within 3 months. Total exon gene detection was also performed and we didn't find any abnormalities. Family history showed that her father had a history of drug abuse. And, no more useful information was found.

JCS is an ill-defined, rare syndrome characterised by axillary freckles, café-au-lait spots and multiple NOFs. JCS has unclear interactions with NF1 till now. The genetic profile of JCS has not yet been determined [1]. It remains intriguing whether JCS is a particular form of NF1 or a separate entity. In the 2013 'WHO Classification of tumours of soft tissue and bone', JCS was defined as the association of NOFs and NF1 [8]. Colby et al. suggested that JCS might be a manifestation of the variability of NF1, as all four patients of their study met the criteria of JCS and NF1 [5]. Stewart et al. also found that in their study, the majority of patients with café-au-lait macules and nonossifying fibromas or giant cell lesions harboured a pathogenic germline NF1 mutation, suggesting that many

✉ Hangzhou Wang
13771843695@163.com.com

[1] Department of Neurosurgery, Children's Hospital of Soochow University, 92 Zhongnan Street, Suzhou, Jiangsu 215006, People's Republic of China

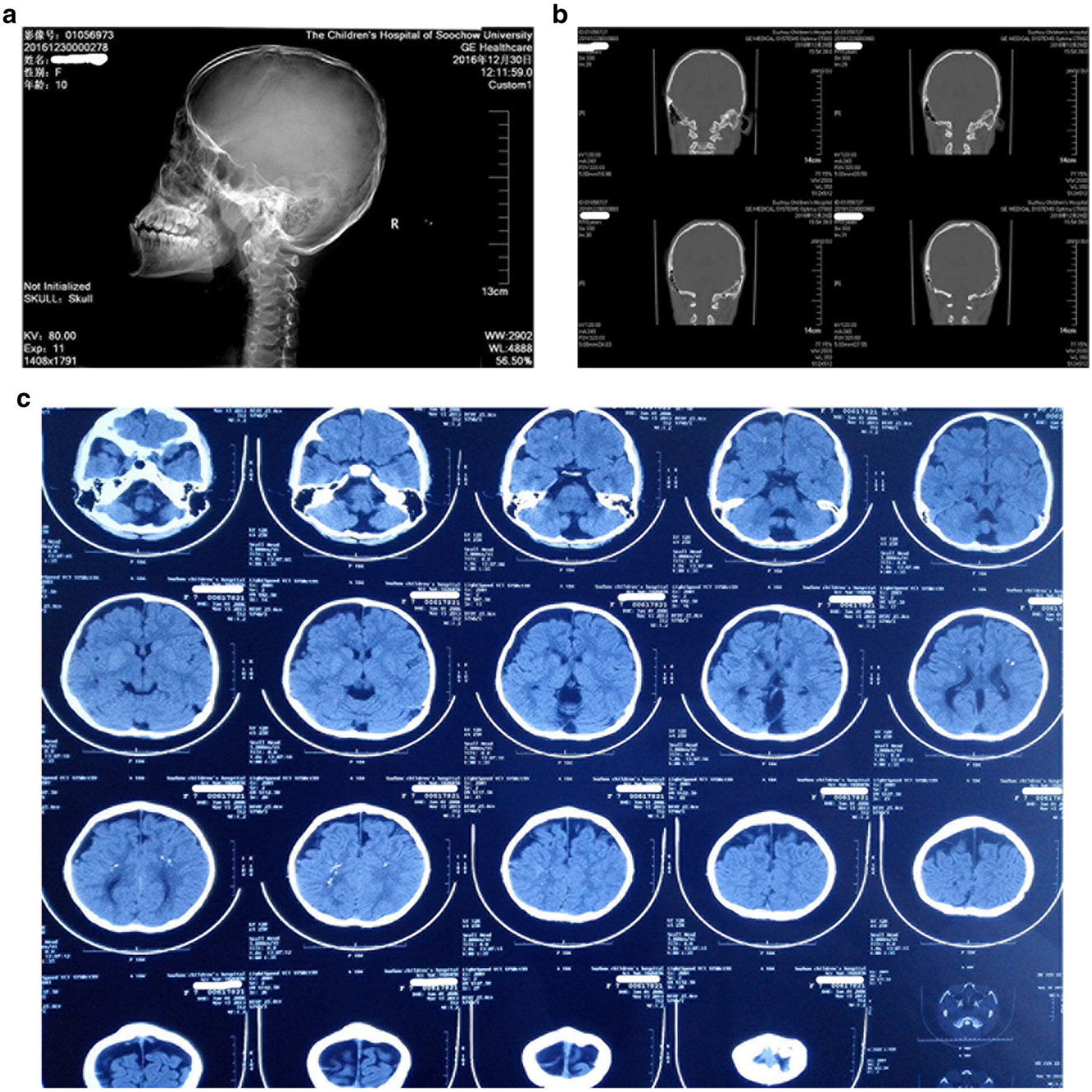

Fig. 1 **a**, **b** Imaging manifestations of a girl with JCS and IADE. The results of X-ray and CT show that the occipital bone was damaged. **c** Sporadic calcification was found in her brain tissue by CT scan

Jaffe-Campanacci syndrome cases may actually have neurofibromatosis type 1 [9]. While in our patient, no abnormality was found by total exon gene detection, and neither skin examination nor whole-body CT scan revealed any evidence of neurofibromas. And this patient met all the criteria of Jaffe-Campanacci syndrome which is proposed by Mirra et al., including axillary freckles, café-au-lait spots and multiple NOFs. We agree with the idea proposed by Colby and Stewart that genetic assessment is critical to define JCS which will help us understand the disease better.

IADE shows an increase in the length and diameter of at least one intracranial artery. The affected intracranial artery extends and sometimes even meandering [10]. The main clinical manifestation of IADE is ischemic stroke [11, 12]. Severe neurological deficiencies can also occur in patients with IADE. Currently, the treatment of IADE depends on the patient's clinical manifestation and the severity of the disease. Concrete methods include blood pressure control, antithrombotic treatment, intravascular therapy and bypass surgery [13–15].

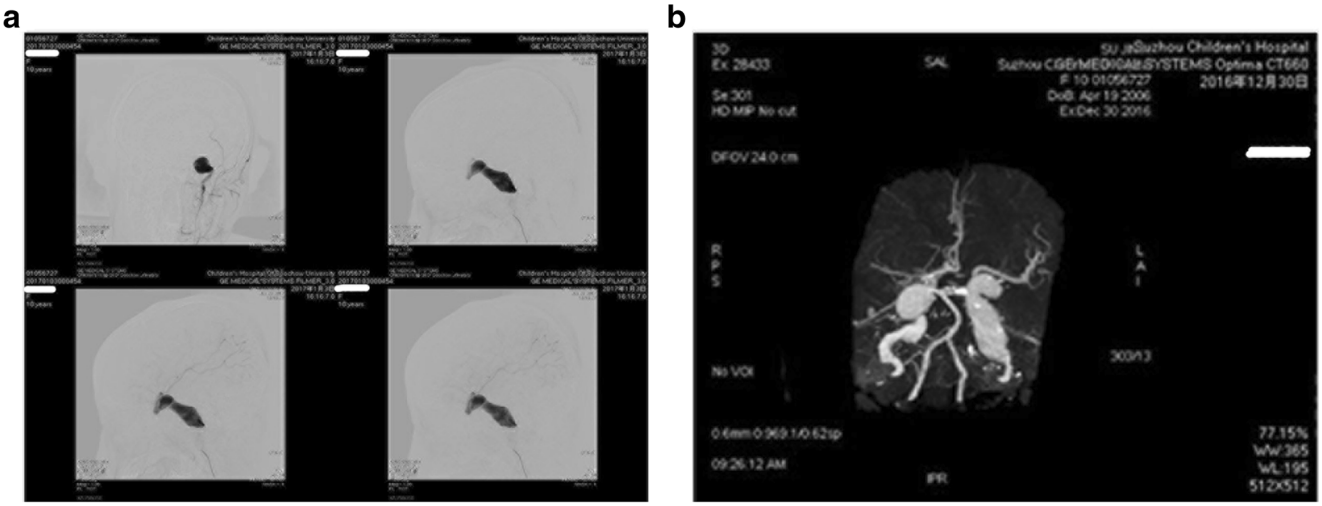

Fig. 2 Intracranial angiography results of a girl with JCS and IADE. **a** DSA and **b** CTA showed that both of the internal carotid artery extended and expanded

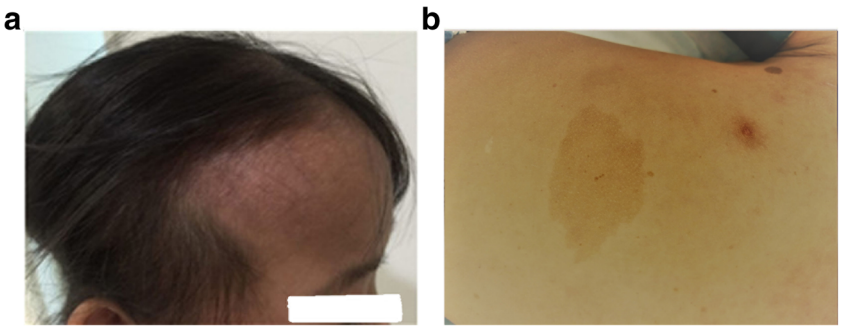

Fig. 3 Morphologic characteristics of a girl with JCS and IADE. **a** Hyperpigmentation with café-au-lait spots is shown on the face of the patient. **b** Axillary freckles was shown

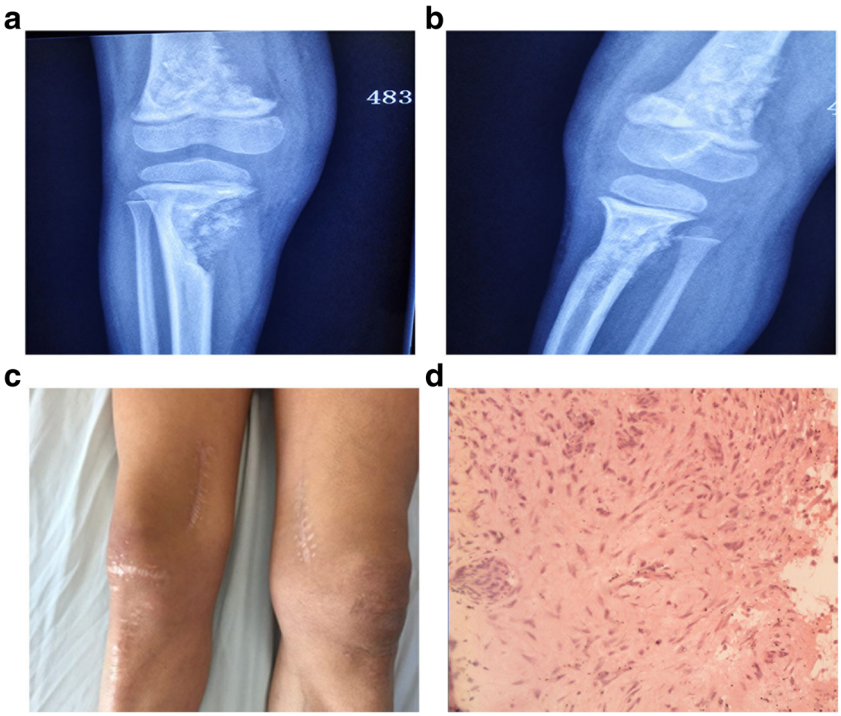

Fig. 4 Bone lesions of a girl with JCS and IADE. **a**, **b** X-ray shows nonossifying fibromas involving the femur and tibia of both sides. **c** Surgical incision of the bone lesions. **d** Bone biopsy suggestive of spindle-shaped fibroblasts in storiform pattern and multinucleated giant cells, typical for NOF (H&E, × 200)

Our patient was a rare example of combined IADE and JCS. In addition, occipital bone damage and multiple calcifications in brain tissue were also observed in this patient. Most reports of IADE or JCS have described only morphologic lesions. And patient with both IADE and JCS has never been reported. The relationship between the two diseases remains to be explored.

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Compliance with ethical standards

Conflict of interest The authors received no financial and/or material support for the research reported in this paper. The authors state that there are no conflicts of interest arising from the research reported in this paper.

Ethics approval and consent to participate Not applicable.

Consent for publication Written informed consent was obtained from the patient for the publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Availability of data and material Not applicable.

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