EYELID COLOBOMA AS A FIRST PRESENTATION OF JAFFE CAMPANACCI SYNDROME

Lilian Emi Ohkawara

Patricia Yuri Miyasato

Midori Hentona Osaki

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Eyelid Coloboma as a first presentation of Jaffe Campanacci Syndrome

Lilian Emi Ohkawara, Patricia Yuri Miyasato, Midori Hentona Osaki
Federal University of São Paulo

INTRODUCTION

Jaffe Campanacci syndrome (JCS) was described in 1982 to describe the complex of multiple no-ossifying fibromas of long bones, giant cell granulomas of the jaw, and café-au-lait macules associated with intellectual disability, precocious puberty, congenital blindness, and kyphoscoliosis. We report a 10-year-old girl with multiple café-au-lait spots, ocular abnormalities, and non-ossifying fibromata of the bones in which the first manifestation was left eyelid coloboma detected just after childbirth.

CASE REPORT

The case is about a girl in which the first clinical manifestation was with upper left eyelid coloboma detected just after childbirth, involving 75% of the eyelid margin associated to conjunctival symblepharon, iris atrophy, microphthalmia, exposition keratopathy, corneal neovascularization, optic disc and inferior retinal coloboma. Superior left eyelid reconstruction was performed with interposition of inguinal skin graft and symblepharon correction, with improvement of the lagophthalmos and the exposition keratopathy. For the microphthalmos the patient was submitted to prosthetic treatment to promote orbital expansion to minimize orbital asymmetry.

During her childhood alopecia, axillary freckles and café-aul-lait macules in the left thorax and abdomen were noted.

At the age 7, the patient had a spontaneous fracture of the left tibia and the computerized tomography showed multiples non ossifying fibromas. At the moment the patient is 10 years old, with visual acuity of 0,9 in the right eye and no light perception in the left eye, on a multidisciplinary follow up. Genetic analysis of NF1, NF2, and SPRED1 was performed, but no genetic variants were found. Despite negative results, it was not possible to discard abnormalities at the non codifying region of the analyzed genes.

PICTURES

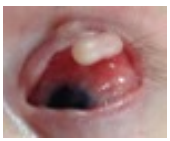
Fig 1: left eyelid coloboma

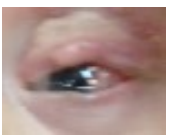
Fig 2: 3 months post operative

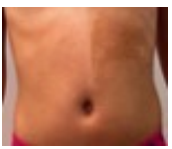
Fig 3: café-au-lait spots

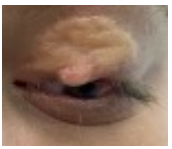
Fig 4: 10 year post operative

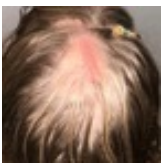
Fig 5: alopecia

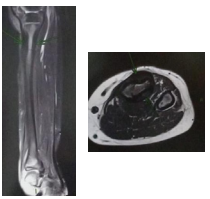
Fig 6 and 7: non ossifying fibromas

DISCUSSION

JCS is a rare disease in which remains intriguing whether is a particular form of Neurofibromatosis type 1 or a separate entity. Despite the inconclusive genetic analysis for JCS, the patient met all the criteria for the syndrome, including axillary freckles, café-au-lait spots and multiples non ossifying neurofibromas. There is no published report in the literature which describes the association of neurofibromas and eyelid coloboma.

REFERENCES

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2. Mankin, Het al: Non-ossifying fibroma, fibrous cortical defect and Jaffe-Campanacci syndrome: a biologic and clinical review. Musculoskelet Surg (2009) 93: 1-7