48th BRAVS Meeting RETINA 2024 CONNECTING SCIENCE TO REAL WORLD

April 18th to 21st
Royal Palm Hall
Campinas - SP
Brazil

COCKAYNE A SYNDROME: A CASE REPORT

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PURPOSE

To report a clinical case about COCKAYNE A Syndrome.

METHODS

Analysis of medical records and imaging.

RESULTS

12 y.o. girl diagnosed with Cockayne syndrome (CS), a rare autosomal recessive disorder characterized by delayed neuropsychomotor development, growth impairment, and progressive neurological dysfunction. At 4y.o., she began follow-up due to developmental delays, with initial fundoscopic examinations showing no abnormalities. By age 6, subtle retinal pigment epithelium (RPE) rarefaction was observed bilaterally. However, fluorescein angiography (FA) was not performed at that time. At 12y.o., CS was diagnosed by a neurologist, prompting a new ophthalmological evaluation revealing early cataracts, increased RPE rarefaction, and slight optic disc pallor bilaterally. FA was performed at 13 y.o., confirming optic disc pallor, diffuse vascular thinning, and pigment dispersion consistent with bone spicules, sugesting retinitis pigmentosa. The patient continues to receive follow-up care from ophthalmology and neurology specialists and is now participating in the "Saber e Ver" Project, funded by the Ministry of Health.

DISCUSSION

CS presents with various manifestations including photodermatitis, growth retardation, and neurological abnormalities such as microcephaly and retinitis pigmentosa. The syndrome progressively leads to blindness, deafness, and death. Diagnosis involves thorough history-taking and physical examination, focusing on neurological, ophthalmological, cutaneous, and dental aspects. Confirmatory tests encompass brain imaging, metabolic screenings, and cerebrospinal fluid analysis, along with assessments of fibroblast sensitivity to UV light and DNA metabolism studies. Ophthalmological evaluations of patients with early-onset retinitis pigmentosa and cataracts aid diagnosis. Management remains symptomatic, with regular assessments, physiotherapy, and sun protection advised.

In conclusion, CS is a rare disorder with severe manifestations. Prompt diagnosis followed by comprehensive multidisciplinary care is crucial to minimize morbidity and mortality impacts.

IMAGES

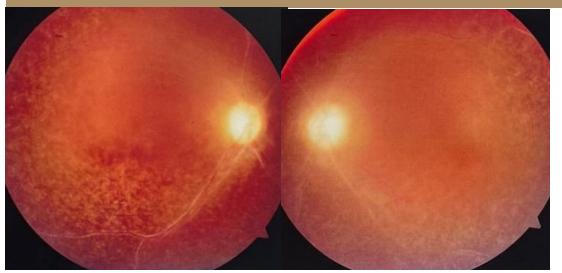

Image 1: Retinography

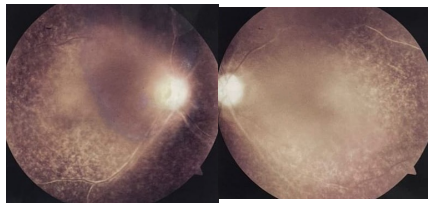

Image 3: Red Free

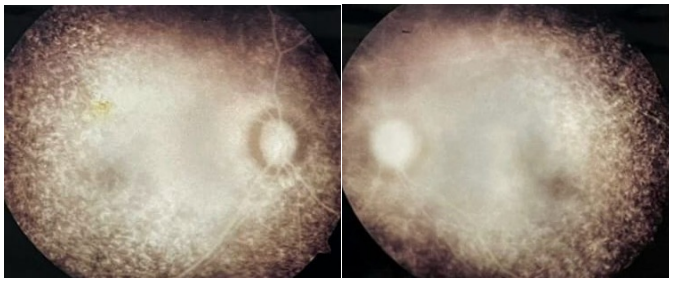

Image 3: FA

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