GENETICS AND GENODERMATOSES

COCKAYNE SYNDROME: ABOUT A FAMILY OF FOUR BROTHERS

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Introduction: Cockayne's syndrome is a rare, multi-systemic, neuro-degenerative, photogenodermatosis of autosomal recessive inheritance. We report four cases of CS in the same family.

Observation: The four brothers came from a first-degree consanguineous marriage, had stunted weight and mental growth retardation, cutaneous hyperpigmentation of exposed photo areas, premature cutaneous aging and sensorineural deafness.The older brother died at the age of 35, because of an AVP. The second 52 years old had a dementia with functional impotence secondary to a significant deformity knee varum with tendon retractions to 4 members. The third brother was 47 years old with asthma, a paresthesia-type neurological disorder with absence of convulsion-free state and cerebellar syndrome, with articular lesions of type of knee osteoarthritis, central and atrophy of the cerebral scan brain. The EEG showed signs of demyelinating neuropathy.
The 4th showed dyskinesia associated with tendinous retractions and on the radiograph of the cervical spine rectitude of the spine with vertebral compression and recoil of the posterior wall. The diagnosis in siblings was suggested by the ophthalmological examination of the mother who had shown unilateral left pigmentary retinitis localized in the peri-papillary region. The ophthalmological examination of the father was normal. The karyotypes of the four brothers and the mother were normal.

Discussion: The originality of our observation lies in the rarity of the CS and the phenotypic variability making the early diagnosis difficult. The diagnostic criteria defined so far are reviewed. These do not appear to be specific or early. The maternal retinitis-type ocular involvement of the mother in a compatible clinical context made it possible to suggest the diagnosis in our case. The evolution in our patients was marked by the gradual onset of neurological complications, which must be diagnosed at an early age, which requires early and multidisciplinary management, involving dermatologists, ophthalmologists and neurologists.