Sensenbrenner syndrome

Sensenbrenner syndrome, also known as cranioectodermal dysplasia (CED), is a genetically heterogeneous ciliopathy, characterized by dysmorphic features including dolichocephaly (with inconstant sagittal craniosynostosis), chronic kidney disease (CKD), hepatic fibrosis, retinitis pigmentosa, and brain abnormalities, with a partial clinical overlap with other ciliopathies.

A retrospective review of four children with Sensenbrenner syndrome treated at the Femme Mère Enfant University Hospital of Lyon from 2005 to 2020 was conducted.

Variants in WDR35 or WDR19 were found in all children. Two of them underwent surgery for a scaphocephaly in the first months of life. All patients developed CKD leading to end-stage renal disease during the first/second decades.

The diagnosis of scaphocephaly may precede the diagnosis of the underlying Sensenbrenner syndrome, thus highlighting the importance of a systematic multidisciplinary assessment and follow-up for craniosynostosis, in order to identify syndromic forms requiring specific management. In Sensenbrenner syndrome, patients' management should be coordinated by multidisciplinary teams of reference centers for rare diseases, with expertise in the management of craniofacial malformations as well as rare skeletal and renal disorders. Indeed, a prompt etiological diagnosis will result in an early diagnosis of multisystemic complications, notably renal involvement, thus improving global prognosis [1)].

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Quinaux T, Custodi V, Putoux A, Bacchetta J, Rossi M, Di Rocco F. Sensenbrenner syndrome: a further challenge in evaluating sagittal synostosis and a need for a multidisciplinary approach. Childs Nerv Syst. 2021 Feb 19. doi: 10.1007/s00381-021-05075-1. Epub ahead of print. PMID: 33606107.