Kallmann syndrome

Kallmann syndrome (KS) is defined by the association of hypogonadotropic hypogonadism and anosmia. It is characterized by a significant clinical and genetic heterogeneity; actually, it may present several non-reproductive non-olfactory anomalies, and all the ways of genetic transmission can be involved in the inheritance of the disease. Although six pathogenesis-related genes have been identified so far, KS remains sporadic in 70 % of the cases, and the genetic diagnosis is not available for all of them.

The purpose of a paper from Massimi et al. is to present a further disease that can enrich the wide spectrum of KS variability, that is cerebral arachnoid cyst.

This 11-year-old boy presented with the typical characteristics of KS together with those related to a sylvian arachnoid cyst. He was admitted because of worsening headache. At the admission, the physical examination revealed eunuchoid aspect, micropenis, previous cryptorchidism, and anosmia. MRI pointed out a large, left sylvian arachnoid cyst, agenesia of the olfactory bulbs/tracts complex, and hypoplasia of the left olfactory sulcus. The child was operated on by endoscopic fenestration of the cyst, followed by transient external drainage for subdural hygroma and microscopic fenestration for recurrence of the cyst. His statural growth is normal but the sexual development still delayed in spite of hormone replacement therapy.

According to the present and the other four cases in the literature, arachnoid cyst should be included among the anomalies possibly accompanying KS date although this association seems to be occasional as far as embryogenesis and physiopathology are concerned [1)].

1)
Massimi L, Izzo A, Paternoster G, Frassanito P, Di Rocco C. Arachnoid cyst: a further anomaly associated with Kallmann syndrome? Childs Nerv Syst. 2016 Sep;32(9):1607-14. doi: 10.1007/s00381-016-3154-7. PubMed PMID: 27379494.