Article: 545
Topic: 69 - Schizophrenia

PARANOID SCHIZOPHRENIA IN KALLMANN SYNDROME: GENETICS AND PSYCHOPATHOLOGY

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Introduction: Kallmann syndrome (KS) is a genetically heterogenous and rare disorder characterized by the combination of hypothalamic hypogonadism and anosmia/hyposmia. In about one third of the patients, mutations have been identified in at least seven different genes. Apart from one published case report, virtually no data are available about possible neuropsychiatric symptoms in KS.
Objectives: Studying the putative relationship between KS and schizophrenia.
Aims: Investigating the genetic etiology in a patient with a clinical diagnosis of KS and paranoid schizophrenia.
Methods: Detailed diagnostic evaluation including genetic analysis of an adult male patient with paranoid schizophrenia, hypogonadism and anosmia.
Results: Psychiatric examination of the 28-years-old male patient disclosed negative psychotic symptoms only, following an acute paranoid schizophrenic episode one year before. He used 15mg aripiprazole daily and testosterone gel 20mg/gr. Hormonal panel showed extremely low levels of LH and FSH. MRI scanning of the brain demonstrated complete absence of the olfactory bulbs. Otolaryngeal examination disclosed complete anosmia. Neuropsychological assessment revealed subaverage intelligence distinctly disconcordant with premorbid functioning, markedly lowered speed of information processing, and impaired executive and social cognitive functions. Genome wide SNP array analysis and mutation analyses for the KAL1, FGFR1, PROK2, PROKR2, FGF8 and CHD7 genes, disclosed no genetic imbalance or any pathogenic mutation.
Conclusions: This first report on a patient with KS and paranoid schizophrenia in whom extensive genetic analyses were performed, stipulates the need for studying a possible increased risk for psychiatric symptoms in patients with KS.

https://doi.org/10.1016/S0924-9338(13)75838-3 Published online by Cambridge University Press