

Delayed puberty and anosmia in CHARGE syndrome: a case report

ABSTRACT

A 26-year-old female presented to the paediatric clinic at 11 years of age with poor growth. The detection of delayed puberty, anosmia, coloboma and hearing impairment led to a diagnosis of CHARGE syndrome. This was confirmed by a heterogenous de novo pathogenic variant c.6955C>T:p.(Arg2319Cys) detected in the CHD7 gene. Detailed assessment, including olfaction, ophthalmic and auditory examination should be part of the evaluation framework in children with delayed growth and puberty.

Keyword: Anosmia; Delayed puberty; CHARGE syndrome