A new clinical presentation of a non-so-rare disease, the Di George syndrome.

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Introduction

DiGeorge's syndrome(DGS), velocardiofacial syndrome(VCFS) and conotruncal anomaly face represent a heterogenous group of disorder sharing a common genetic basis: deletion in chromosome 22q11.2. In DGS, this deletion is present in 90% of cases and rarely in TBX1. Prevalence of DGS/VCFS is 1/4000 births. The classical triad in DGS is conotruncal cardiac anomalies, hypoplastic thymus, and hypocalcemia. However, the phenotypic features are much more variable.

Observation

We report a 30 years old male (height: 165cm) with repeated epileptic seizures associated with severe and severe hypocalcemia. At first, we diagnosed idiopathic primary hypoparathyroidism and started appropriate therapy. Genetic test showed a deletion in TBX1x1. He didn't show more epileptic seizure, but referred chronic fatigue. A complete hormonal balance showed low levels of IGF-1(67ng/ml) and growth hormone deficiency (GHD)(peak: 0,27ng/ml), confirmed after glucagon test. Pituitary gland was normal at MRI. Interestingly, our patient did not present typical mental retardation seen in 22q11.2 deletion. One daughter of our patient died in utero from cardiac malformation

Discussion

In DGS children, short stature occurs in 36% to 67%. Out of these, only 32% have GHD. In adults, only 10% were below their predicted height, but no one with GHD. Mechanisms underlying GH axis defect and/or short stature are not known. Mutations in TBX1 have been reported in five patients. In all cases, no GH deficiency or short stature has been reported.

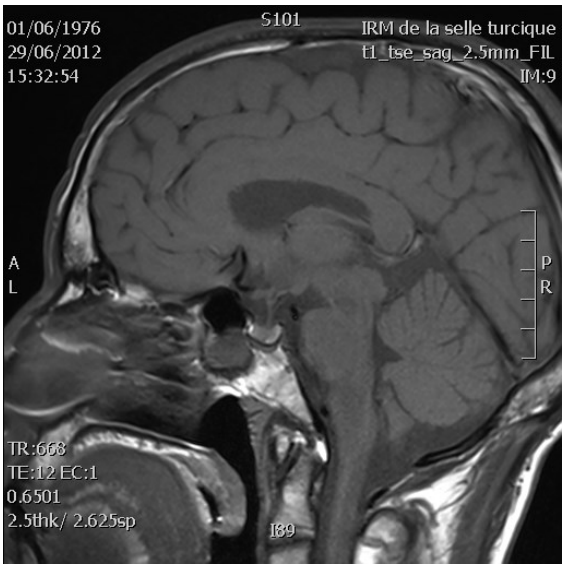

Fig. 1: normal pituitary MRI

Conclusion

We reported here the first case of adult GHD associated with deletion in TBX1. Moreover, our case report confirms the heterogenous clinical presentation in DGS.