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(CASE REPORT)

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Di George syndrome revealed by hypocalcemia: A case report

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Abstract

Di George syndrome or (the 22q11.2 deletion syndrome) is due to a chromosomal abnormality namely a genetic abnormality caused by a microdeletion of chromosome 22. The main manifestations are cardiac congenital malformations, thymic hypoplasia, and hypoparathyroidism with hypocalcemia, psychomotor retardation, craniofacial dysmorphies and sometimes immunodeficiency. The diagnosis can be made antenatally, during fetopathological examination or after birth. We report the case of a 49-year-old female patient who was initially followed for iatrogenic hypoparathyroidism under Levothyrox replacement therapy and whose evolution was marked by the discovery of a Di George syndrome during follow-up.

Keywords: Di George syndrome; Microdeletion; Hypocalcemia; Hypoparathyroidism

1. Introduction

Di George syndrome was described in 1965 by Angelo Di George [1] and combines a characteristic facial dysmorphia, thymic agenesis, absence of parathyroids leading to hypocalcemia, and conotruncal cardiac malformations. It is also called Shprintzen cardiovelo-facial syndrome (cleft palate, heart disease, facial dysmorphia) or Cayler cardiofacial syndrome (heart disease, crying asymmetry). The spectrum of this syndrome is broad with a large number of developmental anomalies described in association with the microdeletion. Di George syndrome is viable: hypocalcemia due to the absence of parathyroids is transient, persisting only in very rare cases; neonatal hypocalcemia is noted in 50% of cases. It usually resolves but may recur at any age or following infection, surgery or pregnancy [2]. Immun deficiency is present in only 1% of cases. Patients with the disease have mental retardation and behavioral problems.

2. Observation

A 49-year-old woman was admitted to the medical emergency room for convulsions related to hypocalcemia with antecedents of muscle cramps, tingling of the upper limbs evolving for 10 years, complicated 6 years ago in post-op of thyroidectomy; by the installation of paresthesias at the level of the upper limbs, inferior, peribuccal, and muscle cramps put under substitutive treatment: levothyrox and calcium.

The clinical examination revealed a dysmorphic syndrome (short neck, low implantation of ears and hair, protruding ears; and dental malformation); the Chvosteck sign was positive. The biological assessment showed: low blood calcium 63.65 mg/l, albumin: 44.3 g/l corrected blood calcium 69 mg/l; phosphorus 38.4g/l, low parathyroid hormone 18.47 pg/mL; low vitamin D: 8.1ng/ml, normal TSH 1.92 IUU/ml. Cardiac MRI revealed a congenital heart disease with not compaction of the left ventricle with flash ETT: FE moderate 45%.

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The diagnosis of Di George syndrome was evoked and the patient benefited from adjustment of her replacement therapy and adaptation of calcium supplementation with treatment for her heart disease.

3. Discussion

Di George syndrome or 22q11.2 deletion syndrome is characterized by a developmental abnormality of the third and fourth branchial arches, which are responsible for the development of the large vessels of the heart, thymus, and parathyroids. Lipson and colleagues reported a cohort of 150 cases of Di George syndrome of which 35% of the cases had a heart defect and 24% a cleft palate, only ten cases had a complete syndrome [3]. Cardiac disease is the most frequent malformation, it is of the conotruncal type [4] in particular: a tetralogy of Fallot, an interruption of the aortic arch with an interventricular or interatrial communication of the ostium secundum type. Conotruncal heart disease is associated with Di George syndrome in 50% [5]. Facial dysmorphia is almost constant but difficult to diagnose because it is usually discreet and may even be absent in African subjects [6]. Hypocalcemia may appear in a variable manner over time and fluctuate throughout life [7]. In 25% of the cases, the patients affected present schizophrenia, which reveals itself in adulthood associated with other psychiatric disorders, most often obsessive-compulsive disorders and depression [8]. The de novo deletion forms are by far the most frequent. There are only 10% of familial forms. A parent with the deletion has a one in two chance of passing it on to his or her offspring, who may express it in varying ways [9]. Each component of the Di George syndrome must be treated for each patient, a detailed evaluation is necessary and a global management must be ensured by the referring physician. The treatment may therefore include hormonal treatment (hypocalcemia - hypoparathyroidism); drugs to improve cardiac function or surgical correction. The follow-up of people with Di George syndrome is multidisciplinary. Psychiatric and neurological disorders may require medication and psychiatric management. Monitoring of blood calcium levels (to adjust calcium supplementation) and thyroid function are necessary throughout life [2].

4. Conclusion

Di George syndrome may manifest itself in adulthood as hypocalcemia. Clinical suspicion should lead to a genetic study. The phenotypic expression of Di George syndrome is very variable, only the knowledge of the different signs of call allows to set the indication of the research of the microdeletion.

Compliance with ethical standards

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Disclosure of conflict of interest

The authors declare no conflict of interests.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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