

UTERUS AGENESIA IN A PATIENT WITH CATCH 22 SYNDROME. PURPOSE OF A CASE

S. Mesonero Cavia¹, S. Maeso Méndez², A. Sarasua Miranda³, I. Díez López⁴.

Residente de pediatría. Hospital Universitario Álava Residente de pediatría. Hospital Universitario Álava
Endocrinología pediátrica. Hospital Universitario Álava Endocrinología pediátrica. Hospital Universitario Álava

INTRODUCTION

The catch 22 syndrome or DiGeorge syndrome or velocardiofacial syndrome, is a frequent pathology with the worldwide prevalence estimated between 1 / 2,000 and 1 / 4,000 live births.

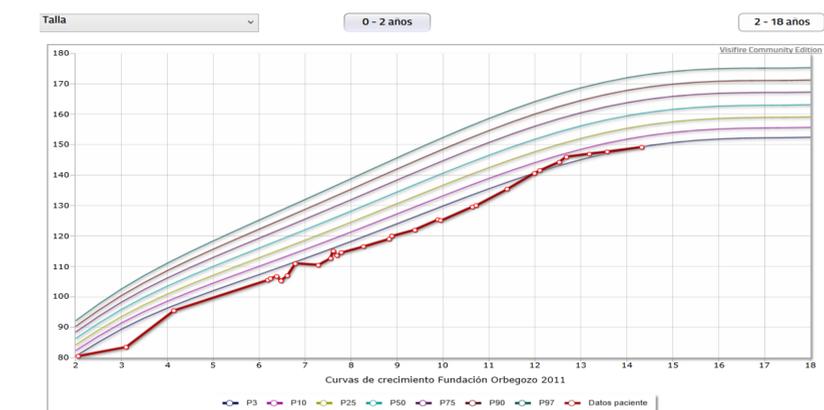
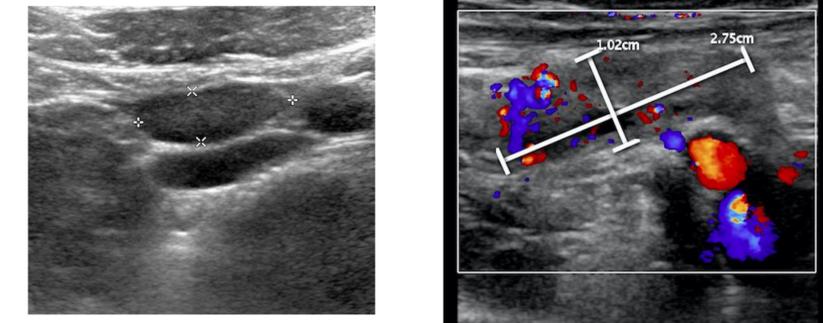
The classic clinic is characterized by typical facial features (narrow palpebral fissures, bulbous nasal tip, small mouth and ears, malar hypoplasia) with functional insufficiency of the soft palate in about 75% of those affected, difficulties in learning, language and behavior, congenital heart malformations in more than 50%, some degree of immunodeficiency (secondary to thymic aplasia / hypoplasia), greater predisposition to develop an autoimmune disease such as hypothyroidism, neonatal hypocalcemia that usually remits with age, although it may recur.

Other manifestations may include gastrointestinal, kidney malformations (multicystic agenesis or dysplasia), or deafness.

However, the phenotype is much more variable and the clinical spectrum continues to expand.

RESULTS

Girl aged 7 years and 6 months at the time of the first child endocrinology consultation, diagnosed with CATCH 22 syndrome. Personal history: Controlled in cardiology due to intervened aortic ring, incomplete right renal duplication in pediatric nephrology follow-up, wearing hearing aids, Velopharyngeal insufficiency in orofacial control by pediatric surgery and neuropaediatric follow-up, normal immunological study without presenting serious infections. On physical examination in the first consultation, she presented Weight: 22Kg, height: 112.6 cm, TA 90 / 60mmHg, with a female velocardiofacial Sd phenotype and tanner stage 1, rest of the normal examination. In the control analysis, no alterations in phospho-calcium metabolism were observed (intact PTH 16 pg / mL [14 - 100], Calcium 9.2 mg / dL [8.4 - 11], Phosphorus 5.1 mg / dL [2.5 - 6]. evolution in consultation starting thelarch at age 11. At age 14 Tanner 4-5 associating several episodes of hypogastric abdominal pain, no menarche. In the event of primary amenorrhea, an abdominal-pelvic ultrasound is requested and a gynecology consultation is observed, revealing: absence of uterus and both ovaries discreetly diminished in size for age.



CONCLUSIONS

In patients with 22q11.2 deletion syndrome, a multidisciplinary follow-up is required. Endocrine manifestations are the most frequent pathology after cardiac ones. From the endocrinological point of view, it is necessary to do a lifelong follow-up, controlling calcemia and thyroid function as the most frequently associated pathologies, but also to take into account the multiple manifestations that may present and be alert to the symptoms of primary amenorrhea since they have described several cases of uterine agenesis in these patients.



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CONTACT INFORMATION

Ignacio Díez López PhD MD
Ignacio.diezlopez@psakidetza.eus

