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Cat Eye Syndrome and Short Stature with Primary Amenorrhea: A Case Report and Review of the Literature

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Abstract: Cat eye syndrome (CES) is a rare disease caused by a chromosomal abnormality with the presence of a small additional chromosome from chromosome 22. It is characterized by a variable association of congenital malformations of which the best known are anal atresia and coloboma of the iris. The colobome, whose appearance recalls the shape of the pupil of the cat, gave its name to the syndrome. Clinical heterogeneity is important. Growth hormone deficiency affects between 1 in 3,000 children and 1 in 4,000 children. Most often, it is idiopathic. Rarely, it is due to a genetic disease or underlying chromosomal abnormality. The association of growth hormone deficiency with Cat-Eye syndrome is very rare. Both a potential link between 22q11 abnormalities and malformations of the uterus has been difficult to determine correctly given the limited case in the literature. We report the case of a patient with growth retardation, primary amenorrhea and a polymalformative syndrome suggestive of Cat-Eye syndrome

Keywords: Cat-Eye syndrome, Growth hormone deficiency, uterine malformations.

INTRODUCTION

Cat-eye syndrome (CES), or Schmid–Fraccaro syndrome (OMIM 115470), is a rare genetic disease with a prevalence estimated between 1 in 50,000 and 1 in 150,000.

This syndrome is known by other names which include chromosome 22 partial tetrasomy (22pter-22q11) or chromosome 22 Inv dup (22pter-22q11) or Schmid-Fraccaro syndrome (OMIM 115470) or chromosome 22 partial trisomy (22pter-22q11) [1]. CES is highly variable in phenotype but criteria for diagnosis include ocular coloboma (of the iris and/or retina), anal atresia (with or without fistula), pre-auricular pits and/or tags, heart defects (especially total anormalous pulmonary venous return), dysmorphic features, such as hypertelorism and down-slanting palpebral fissures, urogenital defectes and mental retardation [2], growth delay is reported in 15% of cases [3]. We report the case of a patient with growth retardation, primary amenorrhea and a polymalformative syndrome suggestive of Cat-Eye syndrome.

CASE PRESENTATION

Patient A.H, 23 years old, admitted for stunting with primary amenorrhea. The examination had found a dysmorphism of the auricular horn and a microcornea, a Tanner: S5P4, a low implantation of the hair and ears. Pelvic MRI: uro-genital malformation associating utero-vaginal atresia and an ectopic pelvic kidney above vesical, presence of 2 bilateral pelvic formations, seat of multiple cystic formations, related to ectopic ovaries. Cerebral MRI: Chiari malformation, Occipital horn dilatation, ophthalmologic examination: bilateral microcorneal, visual acuity: OD: 0.05 OG: 0.05, 47XX karyotype, supernumerary chromosome marker on all mitoses observed and analyzed, the appearance of which is in favor of a supernumerary chromosome marker derived from chromosome 22 (SMCs 22), chromosome-specific chromosome painting FISH is strongly recommended to confirm the diagnosis. Hormonal status: FSH:6.4 IU/ L LH: 4.2 IU / L Estradiol: 17.7 ng / ml, Prolactin: 11.51 ng / ml, cortisol: 23 µg / dl TSH: 4.26 mIU / L, the cardiological exploration did not show abnormalities, the audiogram is in progress, the radiograph of the spine: thoracic kyphosis. Cat Eye syndrome was selected on the basis of karyotype, clinical and radiological features, and malformation table, but utero-vaginal atresia could not be used for hormone replacement therapy prior to vaginoplasty surgery.

Fig-1: Iris coloboma observed in our patient

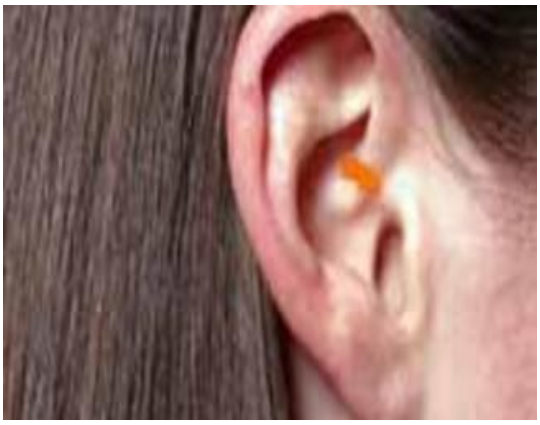

Fig-2: auricular dysmorphism observed in our patient

DISCUSSION

Cat eye syndrome is a chromosome abnormality that affects many different parts of the body. The signs and symptoms of the condition vary widely but may include abnormalities of the eyes, ears, anal region, heart and/or kidney [4-5]. In people affected by cat eye syndrome, each cell has at least one small extra (duplicate) chromosome made up of genetic material from chromosome 22. This extra genetic material leads to the characteristic signs and symptoms of the condition [6]. Most cases of cat eye syndrome occur sporadically in people with no family history of the condition. Cat eye syndrome is most often caused by a chromosome abnormality called an inverted duplicated 22. In people with this condition, each cell has at least one small extra (duplicate) chromosome made up of genetic material from chromosome 22. This extra genetic material leads to the characteristic signs and symptoms of cat eye syndrome [6]. Collectively defects of the urinary system and the reproductive organs are classified as urogenital defects. These two systems have a common origin in the embryo. Typical kidney defects associated with CES include underdevelopment of one or both kidneys (unilateral or bilateral renal hypoplasia); absence of a kidney (unilateral agenesis). Typical reproductive tract defects in females include underdevelopment of the uterus, absence of the vagina or abnormal genitalia. In males defects include undescended testes (cryptorchidism) and external genital abnormalities. Although Müllerian agenesis is the second most common cause of primary amenorrhea the underlying etiology in the majority of cases is unknown. Müllerian agenesis has been reported as a rare finding associated with chromosomal aberrations of the 22q11 chromosomal region including at least one individual with cat eye syndrome (CES) and 10 individuals with deletions or duplications of the 22q11.2 region. However, a potential link between 22q11 abnormalities and uterine malformations has been difficult to adequately ascertain given the limited case reports in the literature. The estimated frequency of short stature in CES is up to 36–50% [3, 7] **,** but GH deficiency has been rarely described in this syndrome. We found two case reports referring patients with CES and GH deficiency [3-7]. In 1975 Pierson et al. describeda child with CES and hypothalamic GH deficiency but failed to dem-onstrate cerebral anomalies. More recently a 16-year-old Saudi male with CES and severe growth failure due to iso-lated GH deficiency was reported [8].

The treatment of CES may require the coordinated efforts of a team of medical professionals. Disease management is directed toward the specific symptoms that are apparent in each individual. In some cases, recommended treatment may also include surgical repair, correction, or management of certain ocular defects, skeletal abnormalities, genital defects, hernias, Hirschsprung disease, biliary atresia, and/or other malformations associated with the disorder. The specific surgical procedures performed may depend upon the size, nature, severity, and/or combination of anatomical abnormalities; their associated symptoms; patient age; and other factors. In addition, individuals with severe short stature in association with growth hormone deficiency may be candidates for growth hormone therapy.

CONCLUSION

In conclusion, this case underlines the importance of acomprehen-sive study of patients with CES, short stature and primary amenorrhea. Subtle pituitary malformations must be looked for. Children with midline defects and short stature should be carefully studied and evaluated for GH defi-ciency, as GH therapy may substantially improve child growth and final height. Additional tests should include thorough eye examination and careful monitoring of hearing. Early recognition of potential visual impairment and/or hearing loss may play an essential role in ensuring prompt intervention and appropriate, early correction or supportive treatment. Specialized imaging techniques and/or other tests may also be used to detect and/or characterize possible gastrointestinal, genitourinary, renal, skeletal, or biliary defects, as well as other physical abnormalities that may occur in association with CES. Investigation of cognitive function may also be appropriate

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