Fraser Syndrome with Major Hydrocephalus

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INTRODUCTION

Fraser syndrome (Cryptophthalmos syndrome) is a rare malformative genetic syndrome described for the first time in 1962 by the British geneticist GR Fraser [1]. Its incidence is 0.043 per 10,000 live births, with autosomal recessive transmission [2].

The authors report a case of Fraser syndrome with major hydrocephalus in order to recall the genetic, pathogenic, clinical and therapeutic aspects of this syndrome.

CASE REPORT

We report the case of a female newborn, hospitalized in Intensive Neonatal Unit, Mohamed VI University Hospital, Marrakesh.

The parents were non-consanguineous; there was a similar case in siblings: a sister who died on the first day of life, having the same facial malformations according to the parents.

The clinical examination had noted a polymalformative syndrome, including: a macrocrania, with a cranial perimeter at 48 cm (>97th percentile), a labiosphenoidal cleft. On the right eye: incomplete cryptophthalmos with the presence of an upper eyelid, without a palpebral fold and without an eyeball. In the left eye: aniridia (fig. 1). No other abnormalities in within the clinical examination except a syndactyly (Fig. 2).

The cerebral CT scan: major hydrocephalus with a reduction of the cerebral parenchyma. (Fig. 3)

The karyotype was normal.

The treatment of our patient required a multidisciplinary approach.

The evolution was fatal after 3 months.

Figure1: cryptophthalmos, labiosphenoidal cleft, macrocrania, agenesis of the nasal pyramid

Figure 2: Syndactyly

Figure 3: The cerebral CT scan: : major hydrocephalus with a reduction of the cerebral parenchyma.

DISCUSSION

Fraser Syndrome (Cryptophthalmos–Syndactyly Syndrome) is a Rare Autosomal Recessive Polymalformative Syndrome;

its Etiopathogenesis is Controversial. It could be due to a primary abnormality of eyelid formation with corneal and conjunctival epithelium metaplasia, a partially formed eyelid refusions- Or due to a failure of programmed cell death that would lead in utero to the opening of temporarily closed areas like: eyelids, fingers, vagina, toes...

Karyotype is normal in this syndrome; recent research has led to the identification of the responsible gene: FRAS1 and research is under way to identify other genes and their homozygous mutations.

About 15% of the children described in the literature are born of consanguineous couples and the history of siblings with this syndrome is described.

Its diagnosis is clinical, with major criteria and minor criteria.

The major criteria are: cryptophthalmos, syndactyly or genital abnormality.

The minor criteria include ear abnormalities, nose abnormalities, larynx and / or palate anomalies, skeletal abnormalities, umbilical hernia, renal agenesis and mental retardation.

The diagnosis is made in the presence of two major criteria and one minor criterion, or a major criterion and four minor criteria.

CONCLUSION

Fraser syndrome remains a major surgical and aesthetic challenge, particularly in developing countries. Need for prenatal diagnosis in order to decide on appropriate management.