Fraser Syndrome in Lubumbashi, Democratic Republic of the Congo: Case Report

Iye Abial Sandra1*, Kintadi Luyingila Ginevra1, Makumyaviri Mbuio Julien1, Nkungwa Kazadi Betty1, Shongo Mick2, Omewatuku Mungomba Jacques-Beaudry3, Yogoole Assani Bienvenu1, Chenge Borasisi Gabielle1

1Ophthalmology Department, University Clinics Lubumbashi, Democratic Republic of the Congo
2Pediatrics Department, University Clinics Lubumbashi, Democratic Republic of the Congo
3Pathology Department, University Clinics Lubumbashi, Democratic Republic of the Congo

Abstract
Fraser syndrome is a rare polymalformative genetic syndrome. It combines major and minor criteria. The diagnosis is made when there are at least two major criteria and one minor criterion, or a major criterion and four minor criteria. It can be isolated or associated with other malformations. The diagnosis is first antenatal and the management is pluridisciplinary.

We report the case of a six-month-old female infant seen at the Lubumbashi University Clinic and whose parents consulted for a congenital malformation of the eyelids of the right eye. The clinical examination noted: complete right cryptophthalmia, bilateral syndactyly (middle and ring) and a broad nose with depression of the ridge. The B-mode ultrasound and the CT scan revealed a bilobed eyeball with antero-posterior diameter lengthening, with the eyeball protruding widely above the bi-ocular line with no crystallinian lens on a redesigned anterior segment.

In this work, we report the clinical and para-clinical aspects of Fraser syndrome for which management remains multidisciplinary and complex. The objective in ophthalmology is to create a palpebral cleft in order to maintain the aesthetic appearance of the face.

Keywords: Fraser syndrome; Cryptophthalmia; Syndactyly

Introduction
Fraser syndrome is an inherited autosomal recessive genetic syndrome [1,2]. Studies had identified a gene called FRAS 1 in 4q21 [3-5]. About 15% of the children described in the literature are born to consanguineous couples [4,6].

It is a rare disease, described for the first time in 1962 [1,7]. The diagnostic criteria proposed by Thomas et al (1986) are clinical. Its main clinical manifestations are: cryptophthalmia, syndactyly, urogenital malformations. The minor criteria are: laryngeal, tracheal, nasal, atrial abnormalities, skeletal abnormalities, umbilical hernia, renal agenesis, and mental retardation. The diagnosis of Fraser syndrome occurs when there are two major and minor criteria, or one major and four minor criteria [4,6,7].

Cryptophthalmia is classified in three stages:
- Complete cryptophthalmia: defined by the presence of a cutaneous sail, which extends from the eyebrow to the cheek, with a total absence of the palpebral fissure.
- Incomplete cryptophthalmia: there is a blank of eyelid and conjunctivaldead end; the cornea is adherent to the skin.
- The congenital syclepharon: there is a syclepharon of the upper eyelid attached to the upper two thirds of the cornea [1,8].

We report the case of a 6-month-old female infant, whose parents consulted at Lubumbashi university clinics for the non-visibility of the right eye since birth.

Method
Our study reports the case of a six-month-old female infant who was admitted in the service for congenital and progressive malformative syndrome of the right eye. The mother reports that the pregnancy was poorly followed (without morphological ultrasound). The child is the seventh child of seven siblings, born from a term pregnancy by a vaginal eutoxic delivery, would have shouted immediately, with a birth weight of 3.150 kg. At birth the mother would have noticed the absence of brow and eyelashes on the right side with the presence of a skin completely covering the right eye without the possibility of opening.

The child fed well and emitted urine and stool without any difficulty. But in the growth of the child, the eyeball seemed to take also volume without notion of pain. This swelling motivated the parents to bring the child for an ophthalmic consultation. The parents testify that there is no family relationship between them and that no child in the family has ever had a similar symptomatology before.

On our examination, the infant presented a good general state and psychomotor development but also:

- Complete right cryptophthalmia (Figure 1): a cutaneous sail
completely covering the eyeball with complete absence of the palpebral fissure;

- Total absence of eyebrow and eyelashes in the right eye (Figure 1);
- A buphthalmia of the same eye (Figure 1);
- A broad nose with depression of its ridge (Figure 2);
- Bilateral syndactyly (Figure 3): fusion of the middle finger and ring finger of both hands.

The syndactyly that the patient presented was a simple syndactyly, where the fusion is purely cutaneous and type B [9].

The B-mode ultrasound (Figure 4) and the CT scan revealed a bilobed eyeball with widening of the unencrypted anteroposterior diameter, the eyeball protruding largely above the bi-acanthal line with no crystalline lens on a remodeled anterior segment. The posterior segment did not present any particularity.

![Figure 1. Complete right cryptophthalmia.](image1)

![Figure 2. Depression and enlargement of the ridge of the nose.](image2)

![Figure 3. Simple bilateral type B syndactyly.](image3)

![Figure 4. Ocular ultrasound in B mode.](image4)

Examination of the left eye revealed no abnormalities. Moreover, the general assessment (clinical, radiologic, abdominopelvic and cardiac ultrasound) with pediatricians and radiologists help, noted no other abnormality (neurology, cardiovascular, renal, locomotor).

**Discussion**

We diagnosed Fraser’s syndrome in view of the following major and minor criteria:

- **Major:** right cryptophthalmia with eyeball remodeled in its entirety, without lens, giving the appearance of a colobomatous cyst, total absence of eyebrow and eyelashes, bilateral syndactyly.
- **Minor:** wide nose and depressed nose ridge.

Several malformations have been described in association with Fraser syndrome, cryptophthalmia being the most recurrent master symptom [10]. Our patient presented a colobomatous, progressive cyst, a form very unusual according to the Slavotinek’s study [5].

The absence of a healthy eyeball on one side and the normality of the left eye and the absence of organic abnormalities on the general level join Atipo [11].

The notion of consanguinity not being found, we find the same results as Amir; as well as the absence of a similar history in the family [12].

Kalaniti, in her study, noted the high rate of death of patients with Fraser syndrome following numerous renal abnormalities [7]. Note that our patient does not have any form of renal damage, her life expectancy remains normal as demonstrated by Meda [6]. In addition, his work has shown that most survivors suffer from severe mental retardation. At 6 months of age, our patient had satisfactory and normal psychomotor development. He seems to escape that too.

All articles published by Research Valley Publishers are Open Access under the Creative Commons Attribution License CC BY-NC.
The management of Fraser syndrome is a function of associated clinical abnormalities and remains multidisciplinary [1,5-7]. The ophthalmological approach is aesthetic for most of the time, consisting of recreating the eyelid cleft.

Antenatal echo-morphology has greatly reduced the incidence of this pathology, making it extremely rare. The severity of the associated malformations makes it possible to make a therapeutic decision as to the progress of the pregnancy [1,12].

Conclusion

Fraser syndrome is a malformation genetic syndrome. The consanguinity incriminated was not found in this case. Thanks to a multidisciplinary examination, our patient has not presented other associated malformations and is fortunate to have a long life.

Declaration of Interests

The authors state that they have no conflict of interest in relation to this article.

References


