

# Neurofibromatosis Type 1 Revealed by Ophthalmologic Complications: A Report of One Case in Ouagadougou, Burkina Faso

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## Abstract

Type 1 neurofibromatosis is an inherited multisystem neurocutaneous disease predisposing to tumors development. Serious skin and ophthalmologic complications, although rare, can occur throughout life. Furthermore in children, unawareness of early symptoms may delay diagnosis. We report the case of A.T. 8 years old, admitted for exophthalmosis and facial deformity dating back to the age of 2 years. The diagnosis of neurofibromatosis was suspected in the presence of light brown skin spots scattered all over the body and subcutaneous nodules. Ophthalmologic examination revealed bilateral exophthalmosis, eyelids neurofibromas, blepharoptosis, Lisch nodules, corneal edema, and optic atrophy. Head CT scan clarified the nature and the extent of ophthalmologic lesions. Treatment was symptomatic. Neurofibromatosis is rarely reported in children in our setting; it is probably under diagnosed. Clinicians should think of this diagnosis in presence of certain specific symptoms and make a clinical assessment.

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## Keywords

Neurofibromatosis, Eye, Complications, Children, Burkina Faso

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## 1. Introduction

Neurofibromatosis (NF) includes two autosomal dominant diseases: neurofibromatosis type 1 (NF1) or Von Recklinghausen disease and neurofibromatosis type 2 [1] [2]. The NF1 is the most common, with an incidence of approximately one case per 3500 births [1] [3] [4] worldwide. It is a genetic autosomal dominant disease,

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whereas *de novo* mutations affect 50% of patients [3]. The mutation of the NF1 genes is located at the pericentromeric region of chromosome 17 [2] [3]. In our context, the rarity of the diagnosis is probably due to ignorance of NF1 clinical signs in children and the delay to seek care.

Clinical signs of NF1 are very diverse and increase in number and size as the individual grows older. The café au lait spots (CLS), visible at birth, are usually the first symptoms. As for eye complications, they can appear anywhere on the eye and at any age [5]. In most cases, symptoms of NF1 are mild, and children live normal life. In some cases, however, NF1 may cause cosmetic and psychological issues. Surgery is often recommended to remove the tumors.

We report a case of neurofibromatosis type 1 in an 8 year old girl revealed by eye complications in the pediatric department of the Yalgado Ouédraogo Teaching Hospital in Ouagadougou, Burkina Faso. Informed consent was obtained from the patient's family to report this case.

## 2. Observation

The eight years old girl was admitted at our department for eyelids blepharoptosis and facial deformity. The onset of symptoms goes back to the age of two years with gradual swelling of the cheeks and protrusion of the eyes. This has led to a series of consultations in their home region.

In October 2012, with the increasing eye protrusion and eyelids blepharoptosis, parents consulted for investigation.

The girl had a history of right upper eyelid tumor since birth. She was not known to have any other diseases other than the current abnormalities. She is second in a family of four siblings. Her growth (weight and height) and psychomotor development were normal. Clinical examination of the biological parents and siblings was unremarkable: no body spots, no deformity. The family had no history of similar diseases and there was no consanguinity between parents.

Examination of the head noted:

- Bilateral eyelid swelling consisting of tender numbs with blepharoptosis more pronounced on the right eye. (**Figure 1**)
- Bilateral exophthalmos (**Figure 1**)
- Hypertrophy of the right hemi face and facial asymmetry
- Bilateral multinodular parotid swelling, compressing ear canals

Examination of the skin showed:

- Light brown spots (café au lait spots), numerous, scattered all over the body, diameter > 5 mm (**Figure 2**).
- Small hyperpigmented freckles (axillary freckles) about 2 to 3 mm in diameter.
- Tumors located on the back, some of them were only palpable, while others seemed to lift the skin above (subcutaneous neurofibromas) as shown on **Figure 3**
- A soft sessile tumor about 3 cm diameter with hypertrichosis (plexiform neurofibroma), located on the back, Ophthalmologic examination gave details of the eye lesions:
- Bilateral plexiform neurofibromas of the upper eyelids
- Lisch nodules (**Figure 4**)



**Figure 1.** Neurofibroma with hypertrichosis on the back.



**Figure 2.** Coffee milk colored spots (café au lait spots). (a) Left arm; (b) left thigh.



**Figure 3.** Parotid swelling + blepharoptosis + exophthalmos of the right eye.



**Figure 4.** Lisch nodule.

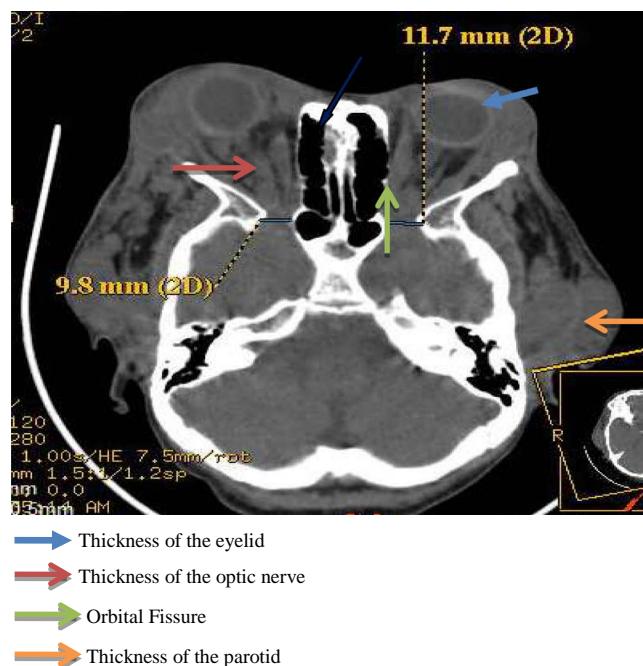
- A thinning of the sclera, corneal edema, and optic atrophy of the right eye. The left eye was healthy.
- The intraocular pressure of the right eye was 41 cm and of the left eye 26 cm Hg (normal value: 10 - 20 cm Hg). The visual acuity was 4/10 on the right eye and 7/10 on the left eye.

Computed Tomography (CT scan) of the skull and brain specified the extent of the lesions (**Figure 5**):

- Left temporal parenchymal calcifications ,
- A bilateral grade III exophthalmos,
- Thickening of the initial portions of the optic nerves and eye muscles

Histological examination of a biopsy of the parotid mass confirmed the diagnosis of NF1. It showed fusiform cell cytoplasm and sometimes undulating pyknotic nucleus with myxoid or "onion bulb" territories compatible with neurofibroma.

Molecular biology in search of genetic anomalies could not be performed.



**Figure 5.** Computed tomography (C.T) scan of the skull and brain.

Treatment was symptomatic consisting of antibiotics, antiseptic and anti-inflammatory eye drops.

The course of the disease was marked by the gradual increase of blepharoptosis, of facial hypertrophy, and of the size of the plexiform neurofibroma on the back. One year later, the patient lost the sight of the right eye.

### 3. Discussion

Diagnosis of neurofibromatosis at a very late stage with severe orbital complications, in our patient, is due to unawareness of the first symptoms of the disease in children.

The café au lait spots are usually the first symptoms of NF. They are often congenital and rarely appear after the age of two [1]-[4]. Our patient had these spots since birth, they were certainly under diagnosed. The spots were scattered all over the body hence the difference with other common skin spots in infants. Literature does precise that they are ubiquity in neurofibromatosis [6].

Neurofibromas rarely appear in early childhood, except plexiform neurofibromas which are often congenital and still can be seen before the age of five years [3]. The dermal or cutaneous neurofibromas appear only during puberty period and are almost always seen in adults [2] [5]. We did not notice any cutaneous neurofibromas in our case this is due to the young age of our patient who was only 8 years old.

Our patient had a lot of subcutaneous neurofibromas scattered on the right hemi face responsible for the hypertrophy. The plexiform neurofibromas, in their important form called “royal tumor” consist of skin and subcutaneous soft swellings with uneven consistency. The overlying skin is often thick and may be the site of hypertrichosis and a brown hyperpigmentation as in our patient. They predominate in the territory of the trigeminal nerve (eyelid and orbital) [2] [3].

There was an important ophthalmologic involvement in our patient. Ophthalmologic manifestations may occur at an early age or later and eyelid disorders are the most common in NF1 [5] [7]. Plexiform neuroma is the classic disorder of the eyelid which is generally unilateral and affects mostly the upper eyelid [5]. The plexiform neuroma in our patient affects both upper eyelids and is marked by a significant distorting blepharoptosis especially on the right eyelid. Blepharoptosis is a common ophthalmologic manifestation of neurofibromatosis and is linked to an eyelid thickening due to the presence of the neurofibromas [7]-[9].

Dysplasia of the sphenoid is the most common cause of exophthalmos in children with enlargement of the orbit [10] [11]. Cases of exophthalmos during meningoencephalocele due to the destruction of the large wing of sphenoid had also been described [11] [12].

Lisch nodules are iris hamartomas which can sometimes be seen with the naked eye. They are pathognomonic of NF1 and are part of the diagnostic criteria [2] [3].

A decrease in visual acuity usually affects the eye on the affected side of the head, sometimes associated with other abnormalities such as deterioration of visual field, or glaucoma [7] [9] [13] [14]. In our patient the loss of visual acuity was associated with bilateral glaucoma and bilateral buphthalmosis.

Other ophthalmologic complications have also been described: thinning of the sclera, optic atrophy [15]. All these ophthalmic signs are probably linked to gliomas of symptomatic optic pathways [15].

Genetic tests performed on the patient and his parents could have showed if this was *de novo* mutation or inherited transmission [16] [17]. These tests are not available in our working environment.

Surgery is the main treatment and must take into account the unpredictability of the disease [8]. This treatment is not yet available in Burkina Faso.

In the end, the course of the disease is classically characterized by deterioration of symptoms during puberty or during an event such as pregnancy. Throughout life various tumors can occur [11]. It is still difficult to predict the prognosis of this long-term illness.

## 4. Conclusion

Clinical diagnosis of NF1 is usually easy. In our study, health workers ignorance of early clinical signs certainly explains the long delay of diagnosis and numerous ophthalmologic manifestations observed. The unforeseeable development of NF1 warrants early diagnosis and regular monitoring throughout life. Molecular diagnostic methods would have determined the etiology of this condition and so consider genetic counseling. This study highlights the need for inter-disciplinary collaboration; without doubt surgery could have improved the quality of life of our patient.

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