

## Case Report

### Late Congenital Leber's Amaurosis in Lubumbashi, Democratic Republic of the Congo: Case Report

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#### Abstract

The authors report a case of congenital Leber's amaurosis on alternate convergent strabismus, left eye fixator discovered in a late stage in an 8-year-old male child at the University Clinics of Lubumbashi. This observation draws the attention of clinical researchers to the need of good early clinical assessment in the presence of retinal dystrophy in children.

**Keywords:** Congenital Leber's amaurosis, Retinal disease, Early onset, Retinal dystrophy, DR Congo

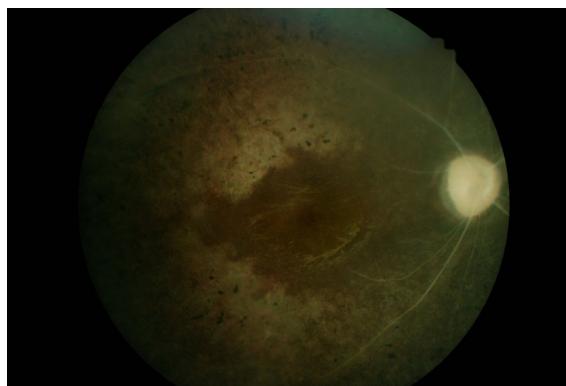
#### Introduction

Described in 1869 by Leber, Congenital Leber's amaurosis is the earliest and most severe form of all hereditary retinal dystrophies. It is an autosomal recessive disease [1]. Its prevalence is between 1/33000 and 1/50000 live births [2]. It accounts for 5% of retinal dystrophies and 20% of causes of blindness among children of school age [3]. It has never been described in the Democratic Republic of the Congo, to our knowledge.

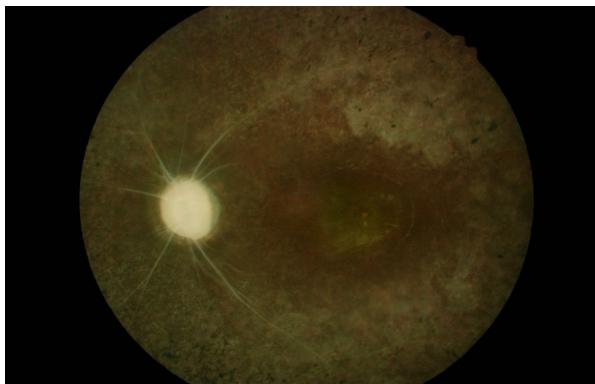
#### Case Report

An 8-year-old boy was brought to the eye clinic for weak vision since the age of one year. Visual acuity was of fingers counting at 4 meters on the right eye and at 5 meters on the left eye and not improvable by corrective lenses on convergent strabismus alternating left eye fixator. Bio-microscopic examination showed a small congenital central cortical opacity on the right eye, and

the anterior and posterior segments were normal on the left eye. Fundus copy of both eyes noticed a macula with fine whitish punctuations having the brilliant appearance of "snail slime", a dark red macula surrounded by greyish retina; optic disc pallor and pigmentary deposits scattered in the periphery of the retinal field and narrowing of the retinal vessels (Figure 1 and Figure 2).



**Figure 1:** Right eye Retinography.



**Figure 2:** Left eye Retinography.

## Discussion

Leber congenital amaurosis is a part of the spectrum of early-onset retinal dystrophy. It usually presents in the first few years of life, most often before the age of 1 year [4].

Leber congenital amaurosis encompasses a group of severe inherited retinal dystrophies responsible for early childhood blindness. There are currently 25 genes implicated in the pathogenesis of these diseases, and identification of disease-causing variants will be required for personalised therapies. Whole exome and whole genome sequencing is informative for detecting novel disease-causing genes, whilst next-generation sequencing has excelled at detecting novel variants in known disease-causing genes. A global effort will be required to identify patient populations for early intervention [5]. Retinal blindness is a major cause of pediatric visual loss. Leber's congenital amaurosis is one of causes of visual loss in children, often wrongly included in the spectrum of retinitis pigmentosa. The disease has become the center of research after initial reports of success in management with gene therapy [6]. Boys are the most affected children (85% of cases); optic disc pallor and peripheral pigment retinitis, macular dysplasia, punctate retinitis, arterial strictures, hyperopia and cataract can be observed (our case), enophthalmos and keratoconus may be also associated ocular conditions. Discovery of the disease at a late stage is rare in the literature [7].

We ruled-out TORCHES syndrome through following laboratory tests described below:

1. Elisa IgG CMV: absence (reference value > 35UI/L)
2. Elisa IgM CMV: absence (reference value < 10UI/L)
3. Elisa IgG Toxoplasma: 135 (reference value > 150UI/L)
4. Elisa IgM Toxoplasma: absence (reference value < 15UI/L)
5. Avidity: absence (reference value < 32UI/L)
6. Elisa Rubella IgG: absence (reference value < 20UI/L)

## Conclusion

Congenital Leber's amaurosis is an inherited congenital retinal dystrophy which leads to low vision or blindness in the first year of life. It can be diagnosed at a late stage of the disease after a good clinical assessment.

**Financial Support:** Nil

**Ethical Aspects**

The patient's parent offered a verbal informed consent to this publication and we preserved his anonymity. The Ethics Committee of the University of Lubumbashi endorsed this study.

**Conflict of Interests**

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

**Statement**

The manuscript has been read and approved by all the authors, the requirements for authorship as stated earlier in this document have been met, and that each author believes that the manuscript represents honest work.

**Declaration of Patient Consent**

The authors certify that they have obtained all appropriate patient consent forms.

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