



Leber's congenital amaurosis. Case report

Amaurosis congénita de Leber. Reporte de caso

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ABSTRACT

Leber's congenital amaurosis is an heterogeneous and genetic clinical disorder characterized by severe loss of vision at birth. It accounts for 10 to 18% of congenital blindness cases. Some patients exhibit solely retinal blindness and show evidence of multi-systemic involvement. The presentation of this case includes bibliographic review of the subject, presentation of a clinical case and description of the importance of stomatologic handling of these patients. Knowledge and understanding of the disease as well as treatment sequels are paramount.

Key words: Leber's congenital amaurosis, congenital blindness.

Palabras clave: Amaurosis congénita de Leber, ceguera congénita.

RESUMEN

La amaurosis congénita de Leber es un desorden clínico, genético y heterogéneo caracterizado por una severa pérdida de la visión al nacimiento. Se presenta en un 10 a 18% de los casos de ceguera congénita. Algunos pacientes muestran solamente ceguera de origen retinal mostrando evidencia de un involucro multisistémico. En la presentación de este caso se hace la revisión bibliográfica del tema, la presentación de un caso clínico y se describe la importancia del manejo estomatológico de estos pacientes, ya que es importante el conocimiento y el entendimiento de la patología y de las consecuencias de su tratamiento.

INTRODUCTION

Leber's congenital amaurosis (LCA) is one of the most severe forms of retinal dystrophies responsible for congenital blindness.¹ It is present in 2-3% out of 100,000 births^{2,3}, and accounts for 10 to 18% of congenital blindness cases reported by Institutes for the Blind,^{4,5} and 5% of all retinal dystrophies. This percentage is probably high in cities where inhabitants exhibit high consanguinity indexes.^{1,6,7}

Many LCA cases present an inherent autosomal recessive pattern. This fact was established by Drs Alstrom and Olson in 1957.^{4,8}

LCA represents a simple disease entity. Some patients only exhibit retina-originated blindness. Nevertheless, others might show evidence of multi-systemic involvement, which can include renal, cardiac, skeletal and especially central nervous system anomalies.

Dr. Harris informed that the first description of LCA was conducted in 1869, when the German ophthalmologist Dr. Theodor von Leber described a congenital disorder characterized by deep loss of vision with the presence of nystagmus, slight papillary reaction and pigmentary retinopathy. Dr. Leber studied a great many cases in a school for blind children. He concluded it was a form of hereditary retinopathy,

where 25% of relatives of affected children presented consanguinity.⁹

Dr. Leber described in those children an apparently normal ocular fundus, exhibiting progressive pigmentation as the children grew up. After this initial Leber's report, it was in 1954 that Drs. Franceschetti and Dieterlé contributed with findings such as strong reduction of null stimulation in the electroretinogram.^{10,11}

To the present date, mutations of six different genes associated to LCA have been identified with LCA: AIPL1, CRB1, CRX, GUCY2D, RPE65, RPGRIP1.¹⁰

Dr. De Laey,¹⁰ proposed in 1991 diagnosis of LCA with the following characteristics:

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- Early blindness or limited vision (first year, before six months of age).
- Slight pupillary reaction.
- Nystagmus/vague ocular movements.
- Ocular digital signs.
- Decreased or absent electro-retinogram.

In addition to these ocular symptoms, another great variety of symptoms can appear such as neurodevelopment delays, mental disability and systemic anomalies.

OBJECTIVE

- To be familiar with clinical characteristics, incidence and etiologic factors of Leber's congenital amaurosis.
- To become cognizant with limitations existing in communication between pedodontist and LCA patients during stomatological rehabilitation.

CASE PRESENTATION

A 7 year 5 month old male patient was referred to the Pedodontics Clinic of the Graduate School, School of Dentistry, National University of Mexico for assessment and treatment of gingival bleeding (*Figure 1A*). The patient came from San Miguel, State of Mexico, and had been previously diagnosed with Leber's congenital amaurosis.

When taking clinical history, personal background of the patient revealed the child was not attending school and cleaned his teeth once a day.

Personal pathological background revealed full term birth at 36 weeks.

Physical examination revealed nystagmus, internal strabismus, slow eye movements, slight papillary reaction, endophthalmitis, positive ocular-digital sign, poor vision, loss of audition, neurological dysfunction, slight mental disability, reduction of muscular activity and poor phoneme articulation (m/a/m/a/ (mother) and a/g/ua/ (water) (*Figure 1B*).

Exploration of the mouth revealed early stage mixed dentition, clinical absence of tooth 62, gingival swelling, presence of dento-bacterial plaque, and multiple grade II and III caries (*Figures 2A and 2B*).

Radiographically, the following could be observed: congenital absence of tooth number 62, presence of all permanent tooth buds; 11,12, 13, 14, 15, 16, 17, 21, 22, 23, 24, 25, 26, 27, 31, 32, 33, 34, 35, 36, 37, 41, 42, 43, 44, 45, 46 and 47 (*Figure 3*).

TREATMENT

Treatment was designed according to the following phases:

First phase: prevention

The patient attended the clinic presenting gingivorrhagia elicited by accumulation of dental plaque in all teeth.

Dental plaque disclosing tablets were used. Tablets had previously been ground and diluted in 5ml water. Tincture was applied with a hyssop (cotton swab).



Figure 1.

A. Clinical characteristics of Leber's congenital amaurosis.
B. Oculo-digital sign in blind patients.

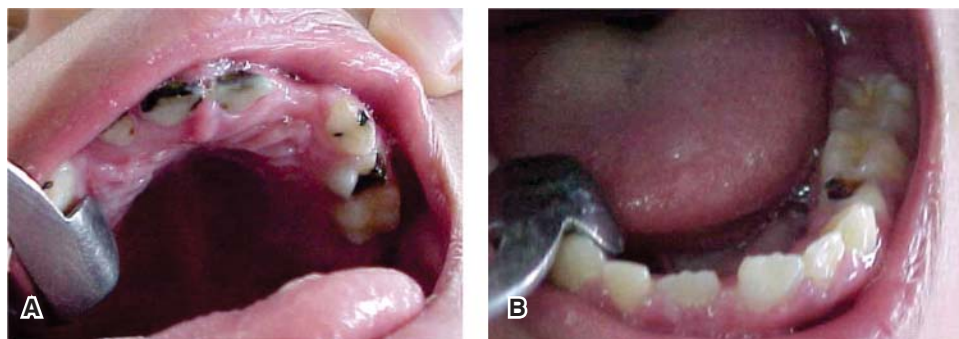


Figure 2.

A. Mouth exploration: grade 2 and 3 caries, congenital absence of tooth number 62.

B. Presence of multiple grade 2 and 3 caries.

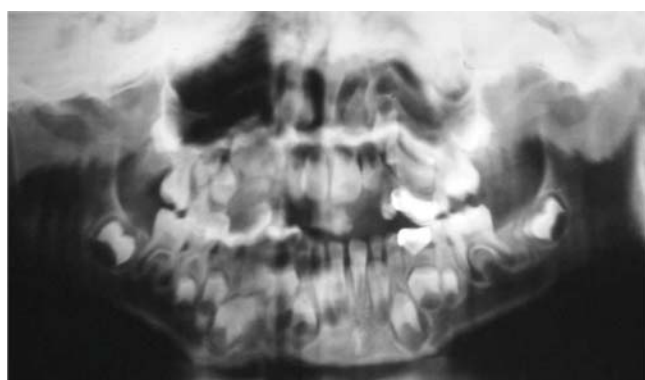


Figure 3. Radiographic evaluation. Congenital absence of tooth 62. Presence of all permanent teeth buds.

The swab was applied to all surfaces of teeth, and the following was achieved:

- First personal plaque control with 100% result.
- Second personal plaque control with 21.4% result.

Teeth were polished with paste and prophylactic brush. Instructions were given to the person responsible for the child's treatment.

Second phase: communication and dental education

- Didactic material: models (*Figure 4A*). This material was used for patients afflicted with visual disabilities. They were made of materials with different textures and hardness degrees, which could resemble a caries process and dental rehabilitation, as well as dental anatomy and periodontal tissues.

Each model presented a label which stated the name of the model as well as explanations in Spanish and Braille.

Practice consisted in presenting didactic materials to the patient so that he could get acquainted with it and could become sensitive to different textures, so as to later relate them to dental, periodontal and caries tissues. Thus it would be easier for the patient to understand the treatment that was going to be given to his mouth, so he could become cooperative and his conduct would take a favorable turn (*Figure 4B*).

Third phase: restoration

Work was undertaken by areas: anterior-superior, upper-right, upper-left, lower- right and lower- left. Two or three teeth in each area were treated and rehabilitated in one session.

First session: Mandibular topic and regional anesthesia was used with total isolation. Treated teeth were number 85 and 84. Teeth were treated with resin and pit and fissure sealers, tooth number 46 was treated with pit and fissure sealer (*Figure 5A*).

Second session: The patient was subjected to mandibular local anesthesia. The following procedures were performed: tooth 74 received a steel-chrome crown, tooth 75 received resin and pit and fissure sealant, tooth 36 received pits and fissure sealant (*Figure 5A*).

Third session: Topical anesthesia was administered to then proceed to infiltrative anesthesia. The area was completely isolated and the following treatments were performed: Tooth 53 received resin restoration, tooth 54 received a steel-chrome crown, tooth 55 received resin restoration (*Figure 5B*).

Fourth session: After using topical anesthesia, the anterior-superior area was infiltrated and isolated with cotton rolls. Treated teeth were 51, 52, and 61. These teeth were about to exfoliate, were already mobile and presented root resorption. Treatment consisted of carious tissue removal, teeth reconstruction with Glass Ionomer Cement and observation (*Figure 5B*).

Fifth session: Topical anesthesia was first used to then proceed with infiltrative anesthesia. The upper left quadrant was anesthetized. Total isolation was observed, rehabilitation was performed in the following fashion: Tooth 63 received resin restoration, tooth 64 received a pulpotomy and chrome-steel crown and tooth 65 received a resin restoration (*Figure 5B*).

In all sessions, dento-alveolar X rays of areas to be treated were taken, as well as digital photographs of the treatment.

Time used for appointments was approximately one hour.

A panoramic X ray was taken upon completion of treatment.

Physical restraint of the patient was used since no communication or control was achieved. This

was due to the fact that the patient presented three different disability types: (sensory, physical and neuro-psychological)

Fourth phase: control

- Third personal plaque control achieving results of 8%
- Cleansing of dyed teeth with brush and prophylactic paste.
- Delivery of cleansing instructions.
- Recommendations: use of stages dental brush, stage 4, Oral-B brand, toothpaste and plaque-disclosing tablets.

DISCUSSION

The patient exhibited a change in conduct after the use of didactic material. This enabled communication between clinician and the patient, who began to show a certain degree of acceptance and cooperation to the dental treatment (*Figure 6*).

Scientific literature illustrates isolated dental treatment for different disabilities (blindness, hearing loss). Nevertheless, no reference has been yet found for patients afflicted with three different types of

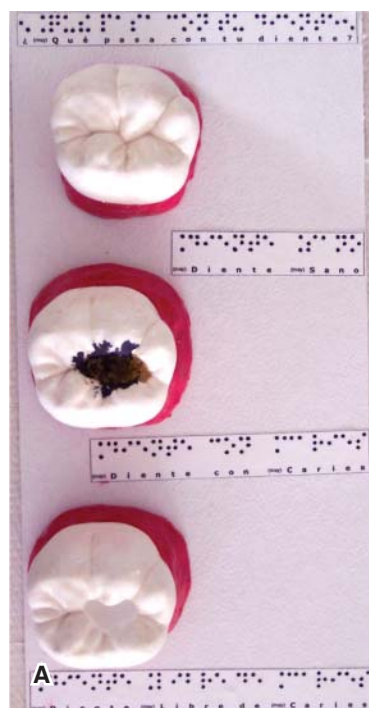


Figure 4.

A. Didactic material used to sensitize the patient.

B. Practices observed to establish communication with the patient.

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Figure 5.

A. Lower oral rehabilitation.

B. Upper oral rehabilitation.



Figure 6. Communication and favorable behavior change were established.

disabilities, with respect to their dental treatment and possible ways of establishing contact with them. No reference has been found of any institution where these patients could be accepted to foster their suitable education, stimulation and treatment.

CONCLUSION

Leber's congenital amaurosis (LCA) is a condition afflicting 2-3:100,000 births. Due to its uncommon occurrence, there is little available information on the subject: why does it appear? How to emit diagnosis? How can I know whether my child is afflicted with this disease? What are its clinical manifestations? At what age can it be diagnosed? Where can I go? Is there any institution which treats afflicted children? Is there any treatment to control the disease? etc.

Dentists, being health promoters, must be aware of the existence of this disease and how to treat it from

the dental perspective. Dentists will then be able to offer specialized treatment which will consist upon dental rehabilitation, prevention of diseases of mouth and teeth, as well as dental education targeted to parents. It is important to deepen the knowledge of the disease in order to be acquainted with possible implications on the dental treatment, recognize the type of disability afflicting the patient so as to establish a suitable treatment plan and offer comprehensive and optimum inter-disciplinary treatment to promote the dental health of patients thus afflicted.

REFERENCES

1. Perrault I, Rozet JM, Greber S et al. Leber congenital amaurosis. *Molec Genet Metab.* 1999; 68: 200-208.
2. Heckenlively JR, Forman SG, Parelhoff ES. Retinal dystrophy and macular coloboma. *Documenta Ophthalmologica.* 1988; 68: 257-271.
3. Leber T. Uber retinitis pigmentosa and angeborene amaurose. *Grafes Arch Klin Exp Ophthalmology.* 1869; 15: 1-25.
4. Schuil J, Meire FM, Delleman JW. Mental retardation in amaurosis congenital of Leber. *Neuropediatrics.* 1998; 29: 294-297.
5. Alstrom CH, Olso OA. Heredoretinopathia congenitalis monohybrida recessive auto Somalis. *Heredities.* 1957; 43: 1-177.
6. Casteels I, Spileers W, Demaerel P et al. Leber congenital amaurosis-differential diagnosis, ophthalmologic and neuroradiological report of 18 patients. *Neuropediatrics.* 1996; 27: 198-193.
7. Foxman SG, Heckenlively JR, Bateman BJ, Wurstschaffer JD. Classification of congenital and early-onset retinitis pigmentosa. *Arch Ophthalmo.* 1985; 103: 1502-1507.
8. Kaplan J, Bonneau D, Frézal J et al. Clinical and genetic heterogeneity in retinitis pigmentosa. *Arch Ophthalmol.* 1990; 85: 635-642.
9. Harris EW. Leber's congenital amaurosis and RPE65. *Int Ophthalmol Clin.* 2001; 41: 73-82.
10. Leroy BP, Dharmaraj S. Leber congenital amaurosis. *Orphanet Encyclopedia.* 2003.
11. Franceschetti A, Dieterlé P. Importance diagnostique et pronostique de l'électrorétinogramme (ERG), dans les dégénérescences tapéto-rétiniennes avec rétrécissement du champ visuel et héméralopie. *Confin Neurol.* 1954; 14: 184-186.

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