

## DENTAL MANAGEMENT OF PATIENTS SUFFERING FROM "LOWE SYNDROME"

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### ABSTRACT

*Lowe syndrome (LS) is an x-linked recessive disorder of unknown etiology resulting in ocular, cerebral and renal disorder. It is Xlinked recessive disorder, caused by a defective gene on Chromosome number Xq 26. Males are more affected than females.*

*Clinical features are bilateral congenital cataract (100%), Glaucoma, Infantile hypotonia, gross motor developmental delay, reduced or absent deep tendon reflexes, muscle wasting, moderate to severe mental retardation, frequent high pitched scream seizures, growth failure, Rickets, Joint hyper mobility and renal manifestations.*

*Prognosis is poor for normal life style. There are developmental delays, visual problems and progressive mental retardation. The patient dies in the first decade of complication of Fanconi syndrome if treatment is not provided. Lifespan can be extended with supportive therapy. A case of LS with dental management, although not ideal is presented.*

*Mentally retarded patients needs regular dental visits to reduce avoidable dental pain.*

**Key words:** *Lowe syndrome, Mentally retarded, Self mutilation*

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### INTRODUCTION

"Lowe syndrome (LS) is an x-linked recessive disorder of unknown etiology resulting in ocular, cerebral and renal disorders." It is also known as Occulo-cerebro-renal Syndrome.

This genetic defect was first described in 1952 by Drs Lowe, Terrey and Mac Lachlan. Incidence is rare (about 50 cases worldwide). Age of onset is new born with hypotonia and cataracts.<sup>1</sup>

It is familial X linked recessive disorder, caused by a defective gene on chromosome number Xq 26 that results in the deficiency of an enzyme called Phosphadidylinositol 4,5-biphosphocataracts. Inzyme is essential for normal metabolic processes that takes place in Golgi apparatus of cell so functions regulated by Golgi apparatus are abnormal, leading to various developmental defects in LS. How the enzyme deficiency leads to these defects is yet not known. Males are more affected than females.<sup>2</sup>

There are three distinct phases of syndrome.<sup>3</sup>

- 1 INFANCY: In which neurologic & ophthalmic manifestations are predominant with renal tubular dysfunction presenting with in first year of life.
- 2 CHILDHOOD: Renal tubular dysfunction, failure to thrive and rickets are common.
- 3 LATE CHILDHOOD: Death from inanition, pneumonia, and chronic renal failure occurs.

### CLINICAL FEATURES

**Ophthalmic Manifestations:** are superficial granulations with corneal scarring, bilateral congenital cataract (100%), Glaucoma +/- buphthalmos, Meiotic pupils, exophthalmoses and visual problems. **Neurological manifestations** are Infantile hypotonia, Gross motor developmental delay, Reduced or absent deep tendon reflexes, Muscle wasting, moderate to severe

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mental retardation, frequent high pitched scream seizures. **Renal manifestations** are minimal at birth but increases in severity with age (usually present with in first year of life) with poly urea & polydipsia. **Other** feature are growth failure, Rickets, Hyperactivity with high-pitched scream, Joint hyper mobility.

### Investigations

- 1 Serum: Low serum bicarbonate, low or normal amino acids, elevated alkaline phosphatase, hypophosphatemia, hypokalemia, hypouricemia
- 2 Urine: Generalized hyperaminoaciduria, glucosuria, organic aciduria, pH <5.5 with low specific gravity, tubular proteinuria, low urinary ammonia, progressive decrease in GFR
- 3 Renal biopsy: Progressive glomerular & interstitial fibrosis.
- 4 Imaging studies: CT/MRI Shows nonspecific abnormalities of CNS.
- 5 EEG: Shows diffuse abnormalities
- 6 X- ray: Shows osteoporosis and rickets

### Management

Prognosis is poor for normal life style. There are developmental delays, visual problems and progressive mental retardation. The patients die in the first decade of complication of Fanconi syndrome if treatment is not provided. Lifespan can be extended with supportive therapy.

### Supportive

- No causative treatment for syndrome
- Multidisciplinary approach
  - a) Pediatric: Vit D supplements, correct metabolic acidosis, hypophosphatemia.
  - b) Neurology: Monitor hypotonia & developmental delay
  - c) Nephrology: Monitor Fanconi syndrome complications
  - d) Ophthalmology: Monitor Ophthalmologic complications
  - e) Genetics: Genetic counseling, prenatal diagnosis (female carrier show fine lenticular opacities on slit lamp examination, greater than 100 opacities in the equatorial area of both lenses).<sup>3</sup>

### Case Report

A nine months old, mentally retarded female patient was brought to the dental department of Children's Hospital PIMS Islamabad on 8th January 2003. Her mother was complaining of lacerated tongue of the

patient due to her newly erupted teeth. According to the mother the patient screamed in high pitch off and on without any reason and self-mutilated her tongue during these seizures.

Upon intra oral examination of the patient only upper primary central and lateral incisors were found, her lower central and lateral incisors were absent. Her mother stated that the patient pulled out her lower teeth during a seizer attack. Tongue of the patient was badly lacerated and infected. Incisors present were of normal colour, size and shape. Teeth were examined for any sharp edge but no sharp edge was found which may cause trauma.

Extra oral examination revealed cataracts in her both eyes (Fig 1). Her mother put on her, woven gloves to prevent the hands from self mutilation, but still fingers of both hands were mutilated and infected (Fig 2). There was an operation scar on her abdomen, which her mother stated was performed for removal of lump when the patient was 3 months old. Decreased muscles tone was also found. No other abnormality was found on physical examination.

Family history revealed that her parents were first cousins. Other sisters and brothers were normal. One of her paternal uncle had two mentally retarded children, one male and one female. Both of them had similar condition as of the presented patient and they died at the age of 10-11 months.

Investigations were performed. Blood serum showed elevated levels of alkaline phosphatase. Urine examination revealed increased amino acids and pro-



Fig 1. Cataract in both eyes of the patient.

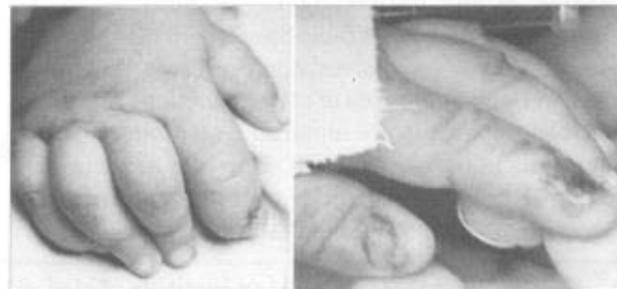


Fig 2. Self mutilated fingers and thumb of both hands.

tein levels. PH of urine was low, i.e., acidic in nature. Her wrist and hands x-rays showed osteo-porosis.

Differential diagnosis of the syndrome was made from the following:

- 1 Occulo dentodigital dysplasia: There is webbing of 4th & 5th fingers, an abnormal small transparent part of the eye, defective enamel & dry hair
- 2 Occulo cerebrorenal syndrome: Characterized by cataract in eyes, renal manifestation and in majority of patient's mental retardation.

On the basis of the clinical picture and laboratory findings the patient was diagnosed with Lowe syndrome.

Counseling of mother was done and mother was advised to give feeder to the patient whenever patient starts screaming. Amoxil syrup and Nilstat drops were given to the patient for infected and lacerated tongue. Patient was referred for dressing of her fingers and recalled for review after two weeks, along with previous medical record.

After a week the patient was brought again to the dental department of children's hospital in emergency around 11 A.M. The patient was screaming in high pitch and fresh blood was oozing from the lacerated tongue. Mother was very upset and reported that the bleeding was started one hour before presentation. She also complained about non-cooperative husband who was not allowing her to bring the patient to hospital for any kind of treatment. Due to continuous bleeding and family problems it was decided to extract all four teeth under general anesthesia immediately. Teeth were extracted and wound was sutured with 3/0 vicryl for hemostasis. Suturing of the tongue was tried but was not possible due to infection.

Patient was medicated and discharged in the evening after full recovery from general anesthesia in stable condition. She was scheduled for review after one week along with previous medical record but she never turned up.

## DISCUSSION

Lowe syndrome is a very rare condition; only few cases had been reported in literature around the world. Three major organ systems involved are eyes; brain and kidneys which are more serious and required more attention than dental problem, so dental management of lowe syndrome was not found in literature.

In the present case, patient was brought to dental OPD due to badly mutilated tongue and fingers with teeth. To prevent further mutilation construction and cementation of splint to cover the incisal edges of upper

teeth was planed. There is always danger of dislodgment of splint in such young and specially mentally retarded patients, but no other treatment option was available. Another problem was construction of another splint as the permanent teeth erupt in future. Poor oral hygiene in mentally retarded patients is quite common and splint may lead to periodontal and fungal infection very easily.

Beside these disadvantages splinting has advantage of retaining natural teeth without causing trauma to tongue during seizer attack. Unfortunately splint was not considered as a treatment option for this patient due to her emergency condition and uncooperative parents.

Patient was at her maternal grandfather's home at the day of emergency and was brought to hospital without her father's permission. After receiving emergency treatment her mother did not report for follow up, probably she was afraid of her husband.

In our society care takers of patients with any mental or physical abnormality usually do not provide correct information. The parents tend to avoid exposing these children to the public or to the hospital, even for necessary treatment. If, by chance they seek medical treatment they usually do not provide correct information to the health care provider. In the present case mother of the patient was complaining of her husband who was rude and was not allowing his wife to take the patient to the hospital or even outside the house. He insisted let the child die. He was not willing to spend money or time on this child. This attitude needs change. These mentally retarded children should be treated with more attention because they are unable to describe their pain, discomfort or problem. The health status of such patients can be improved by counseling of their parents and with supportive treatment to make their life somewhat easier.

The parents of such patients should be encouraged through media to bring these special children for medical treatment. Moreover, Government should provide special facilities in hospitals for such mentally handicapped patients so that they could get special attention and treatment on priority basis.

## REFERENCES

- (LS is a very rare condition and only few cases had been reported in literature around the world)*
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