

## Unusual Enamel Hypoplasia Associated with Teeth Mobility in a 13 Year Old Girl with Wilson Disease

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### Abstract

Wilson disease is an autosomal recessive disorder caused by mutations in the ATP7B gene. It is characterized by the progressive accumulation of copper in the body leading to liver cirrhosis and neuropsychological deterioration. This case may be the first one reported Wilson disease in association with remarkable enamel hypoplasia and teeth mobility leading to severe teeth destruction and pulp exposure. The objective of this investigation was to introduce the dental management for a 13 year old female patient with Wilson disease. The patients restored her smile and she was highly satisfied of the dental work. In conclusion, the dental management of patients with Wilson disease should become the focus of research because of the difficulty in patients' management as our patient was suffering from dystonia restricting the mouth opening and in addition of being a mouth breather which affected the time and quality of the dental work.

**Keywords:** Wilson disease; Enamel hypoplasia; Periodontal disease; Copper disorder metabolism

### Introduction

Wilson's disease (WD) is an autosomal recessive disorder caused by mutations in the ATP7B gene. It is characterized by the accumulation of copper in the body leading to liver cirrhosis and neuropsychological deterioration including movement disorders or dystonia. Kayser-Fleischer rings in the cornea and low serum copper are important keys for diagnosis. D-penicillamine, and zinc salts are known therapies for WD [1,2].

The objective was to introduce dental management to a patient having severe enamel hypoplasia and teeth mobility. Up to the available literature, this is the first case that correlates the WD with unusual dental findings.

### Case Report

A 13-year-old girl was referred to the Oro-Dental Genetics Department, National Research Centre, Egypt. She was diagnosed as WD under the basis that she has a neurological affection including movement disorders represented by poor coordination, loss of fine-motor control, cramped handwriting. Rigid dystonia represented by rigidity and gait disturbance. Pseudobulbar symptoms represented by dysarthria, drooling, and difficulty in swallowing. Psychiatric affection showed depression, aggressive/anti-social behavior and emotional lability. Kayser-Fleischer rings are apparent by direct visual examination. Laboratory investigations revealed low serum ceruloplasmin (40 mg/litre) and elevated basal 24-hour urinary excretion of copper (400 microgram/24 h). Liver biopsy revealed steatosis, glycogenated nuclei in hepatocytes, focal hepatocellular necrosis, fibrosis or cirrhosis and increased hepatic parenchymal copper concentration but she was in a chronic liver state. No bleeding was recorded. Magnetic Resonance Imaging (MRI) of the brain revealed hyperintensity on T2 MRI in the region of the basal ganglia, thalamus, and brain stem. The condition was progressive till she was diagnosed and started treatment. The patient received treatment in the form of D-penicillamine, Zinc in addition to diet control by avoiding foods with very high concentrations of copper.

An extraoral examination revealed synophrus, antimongoloid slanting, slight ptosis, hirsutism, thick lips, everted lower lip, fissured

lips and prominent philtrum (Figure 1a). Intraoral examination showed high arched palate, anterior open bite and enamel hypoplasia (Figures 1b and c). High caries index and pulp exposure were noticed in the case.

Generalized mobility was observed in the whole set of teeth. Dental diagnosis was performed for the siblings and no teeth abnormalities were found. A treatment plan was proposed to restore the aesthetics and function.

Local anaesthesia (mepecaïne-L, Mepivacaine HCl 2%, Levonordefrin 1:20000) was introduced to the patient.

Root canal treatment for anterior and premolar teeth was performed in a single visit, while the molars were finished in three visits. Ready-made posts (Dental Gold Plated Screw Posts, Nordin; Swiss) were used followed by composite resin restoration (Figure 2).

Full mouth rehabilitation was done by using three unit porcelain fused to metal bridges for restoring aesthetic and occlusion (Figures 3a and 3b).



**Figure 1a:** Synophrus, antimongoloid slanting, ptosis, hirsutism, thick fissured lips, everted lower lip, & prominent philtrum. **1b:** High arched palate. **1c:** Open bite & enamel hypoplasia.

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Figure 2: Root canal treatment, ready-made post & composite restoration.



Figure 3: a, b Full mouth rehabilitation.

The patient and her parent were instructed to follow a dental home care regimen. A complete year follow up was done; the patient was highly satisfied by the dental work.

## Discussion

Up to the available literature, this case is thought to be the first one; reported unusual enamel hypoplasia in WD.

In our investigated case, the enamel hypoplasia and the high caries index may be attributed to the presence of the disordered copper metabolism. A strong relationship between enamel hypoplasia and the spread of dental caries was proved [3]. The high caries could be related to the loss of the antimicrobial effect of the copper presented in the patient's saliva due to improper copper metabolism either present in her saliva or in her serum. The copper has an antimicrobial effect on cariogenic bacteria and inhibits the dissolution of the human enamel so prevents demineralization directly as detected [4]. The limited mouth opening and the dystonia showed in this case could also explain high caries lesions as the patient is not able to brush her teeth and to improve a good dental home care. The presence of joint involvement, joint pain and dystonia may be a symptom of WD [2].

The presence of bone affection and periodontal diseases present in our investigated case represented by grade 1 tooth mobility may be explained as copper plays a role in synthesis and cross linking of elastic and collagen. Excessive accumulation of copper in the tissues will give rise to various signs and symptoms [1]. The tooth and its periodontal apparatus including periodontal ligament and bone are formed of collagen [5]. Low copper serum level affected thickness of bone matrix and osteoformation [6].

The periodontal disease detected in this case may be due to the

increased level of copper in the saliva. The elevated level of copper in connection with zinc deficiency in gingiva causes the increase of permeability of gingival epithelium for bacteria. The stimulated inflammatory infiltrate produces more IL-1 and leads to periodontal diseases as reported [7].

Our patient is under D-penicillamine treatment which may exhibit features of damaged elastic fibers in the mucosa and periodontal apparatus [8].

Patient's age, dystonia, and mouth breathing should be considered in our treatment plan decision so fixed appliances were preferred to removable ones. Three unit bridges were constructed to restore severely destructed teeth and act as mobile teeth splint according to Fardal and Linden [9].

Regarding dental management of investigated case, amide group of local anesthesia was used as her liver was not impaired.

Mouth breathing, difficulty in swallowing, deep breath and salivation made long standing mouth opening uneasy in this patient. The patient with WD has an autonomic involvement due to neurological affection as reported [6]. The patient was totally uncooperative to introduce periapical X-ray film to determine working length during root canal treatment which affected the quality of the dental work.

## Conclusion

Oro-dental management of WD is mandatory as early as possible. Oral hygiene care is very important for both patient and parent with longer follow up. Oro-dental aspects regarding WD need more studies. Usage of local anesthetic type is chosen according to liver condition. General anesthesia is more recommended than local anesthesia when the disease is in its chronic phase. Apex locator for working length determination is more advisable than routine periapical X-ray.

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