CASE REPORT

DENTAL REHABILITATION AND CONSIDERATIONS IN PATIENT WITH ABETALIPOPROTEINEMIA : A RARE CASE REPORT

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ABSTRACT

Abetalipoproteinemia is an autosomal recessive disease resulting from mutations in the gene encoding microsomal triglyceride transfer protein. Affected patients present with a wide range of clinical symptoms during infancy. Typical manifestations are failure to thrive, low level of cholesterol and fat malabsorption. Other features like fatty liver, acanthocytosis and anemia are usually present. Such individuals with compromised immunity may be especially vulnerable to the effects of oral diseases. Children affected with more severe conditions have increased risk of having unmet dental needs. Here we present a rare case report of a 5-year-old male with abetalipoproteinemia with multiple carious teeth managed with behavioural management techniques.

KEY WORDS

Behaviour management; Malabsorption; Phenotype; Triglycerides.

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INTRODUCTION

Abetalipoproteinemia (ABL) also known as Bassen–Kornzweig Syndrome is a rare metabolic autosomal recessive disorder caused by mutations in the microsomal triglyceride transfer protein (MTP) geneleading to markedly low levels of triglycerides in plasma along with low cholesterol and fat-soluble vitamins. The prevalence ranges between less than 1 in 100,000 and less than 1 in one million.[1,2]

The syndrome was first described by Bassen and Kornzweig in 1950. In 1958, Jampel and Falls observed low serum cholesterol values in affected individuals and Salt noticed the absence of serum beta-lipoprotein in a patient with this syndromein 1960. Consequently, the name of the disease was later changed to ABL.[3,4]

Children are usually asymptomatic at birth but develop digestive symptoms in early childhood such as intestinal fat malabsorption and failure to thrive. In later childhood or adolescence, ABL patients may develop ataxic neuropathy and retinopathy. Early diagnosis and management of ABL patients is important in preventing manifestations of fat-soluble vitamin deficiencies.[1,5]

Here we present a rare case report of a pediatric patient with abetalipoproteinemia with multiple carious teeth managed with behavioural management techniques.

CASE REPORT

A 5-year-old male patient reported to the department of pedodontics and preventive dentistry with the chief complaint of pain in right upper back tooth region and inability to eat for the past one week. The medical history was significant with failure to thrive at the age of 2-3 months of age. The patient's parents also gave history of large volumes of watery, foul smelling, sticking to the pan and greasy stools. Developmental history revealed slight slurring of speech. Family history revealed consanguineous marriage of the patient's parents.

Extraoral examination revealed a straight facial profile with maxilla and mandible in a harmonious relation. On examining intraorally, multiple carious

PREOPERATIVE DENTITION PICS

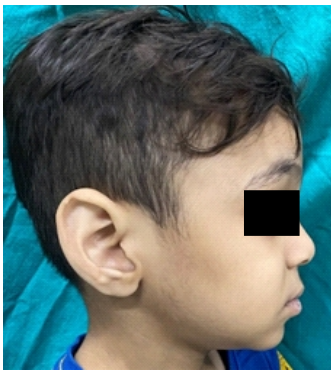

FIG 1: LATERAL FACIAL PROFILE

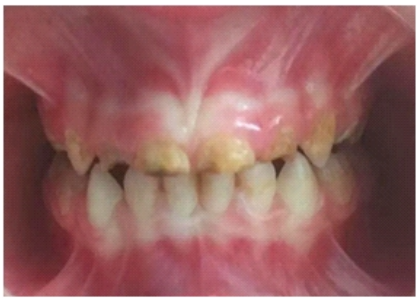

FIG 2

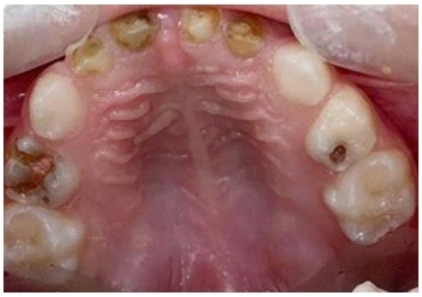

FIG 3

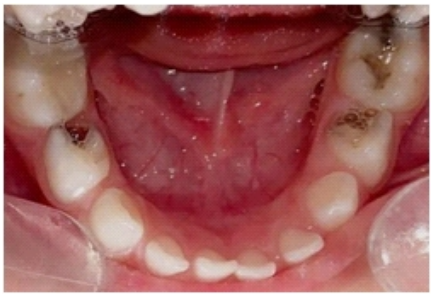

FIG 4

teeth involving 51,52,53,54,61,62,63,64,74,84were observed. There was normal overbite and overjet with end on molar relationship. (fig 1,fig 2, fig 3, fig 4).

The patient was subjected to various radiological and hemotological investigations. Radiographic examination included an orthopantomogram (OPG) (fig 5) and intra oral periapical radiograph. OPG was suggestive of normal tooth development and presence of developing permanent teeth buds as expected of the dental age. Ultrasonography of abdomen and upper gastro-intestinal endoscopy revealed mosaic pattern of the descending duodenum mucosa with oedematous villi and snow-flakes like appearance occasionally. Total cholesterol and triglyceride levels was documented to be <50% mg and <30% mg on more than two occasions. Peripheral blood smear was suggestive of a canthosis (50-60% of RBC's). Based on history, clinical, radiological and hematological investigations, final diagnosis of abetalipoproteinemia was considered.

The concerned paediatrician was consulted before starting the dental procedure. Measures were taken to avoid the stress to avoid any mishaps during the treatment procedure. In the present case, the child was cooperative and managed using non-pharmacological behaviour management techniques

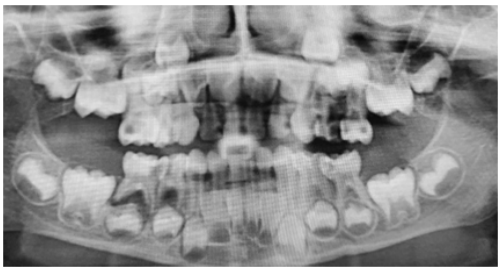

Fig 5: OPG Revealing The Dentition Status

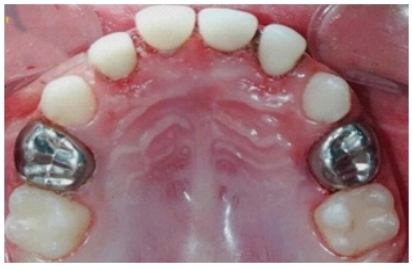

Fig 6: Postoperative Maxillary Occlusal View

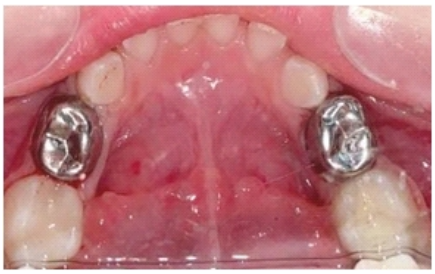

Fig 7: Postoperative Mandibular Occlusal View

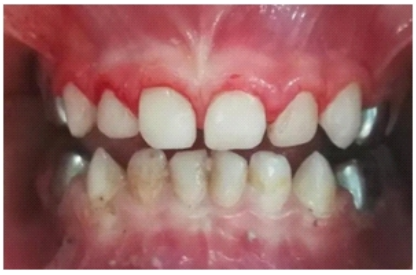

Fig 8: Immediate After Anterior Preformed Zirconia Crown Placement

such as systematic desensitisation, tell-show-do technique and live modelling. All attempts were done to keep the treatment span as short as possible. Carious lesion involving 51,52,61,62 was restored via preformed zirconia crowns; 53,63 restored via composite resin; 74,84 restored via stainless crown under local anaesthesia. (fig 6, fig 7, fig 8).The patient was kept on periodic follow up and encouraged to maintain oral hygiene. There was no further worsening of oral or systemic signs or symptoms noted.

DISCUSSION

ABL is a very rare recessive metabolic disorder characterized by the absence of apoB-containing lipoproteins in plasma. Malabsorption of fat and fat-soluble vitamins leads to variable clinical phenotype that presents in early childhood with steatorrhea and failure to thrive and may include progressive multi-system abnormalities as the patient ages.[6] Most patients are diagnosed in the 2nd to 4th decades and few in the 1st and 6th decades. Earlier presentation of symptoms may be due to a more severe phenotype and may be more resistant to medical treatment resulting in poor outcomes. On the other hand, later presentation and longer period of untreated disease may be associated with poor outcomes due to consequences of fat-soluble vitamins deficiency.[4] Our case was presented in the 1st decade of life.

Consanguineous marriages are associated with an increased risk for congenital malformations and autosomal recessive diseases, with some resultant increased postnatal mortality in the offspring of first cousin couples.[7] A novel non-conservative missense mutation (H529R) was accountable for ABL phenotype in a six-month-old Iranian female infant which was born to consanguineous parents.[8] The present case also revealed consanguineous marriage of the patient's parents.

In ABL patients, the fat-soluble vitamin especially vitamin E reduces markedly though vitamins A, D, and K levels are not so significantly affected. This could be due to the fact that vitamin E predominantly relies on Apo-B mediated intestinal absorption. Most of the vitamin E in intestinal cells gets incorporated into chylomicrons. However, vitamin D is not dependent on lipoproteins for their absorption although vitamin A and K initially follow the journey through intestine and liver via the conventional lipoprotein way, subsequently they have their independent transportation systems in the circulation. Vitamin E deficiency is a chief factor in the pathogenesis of ABL. In patients with vitamin E deficiency, lipoperoxidation leads to the formation of pigment and these deposits in multiple organ tissues like skeletal striated muscle, liver, myocardium, spinal cord, even involving neurological system.[2,9]

Although few ABL patients have had developmental delays or intellectual disabilities intelligence is usually unaffected in ABL. Similarly, in the aforementioned case there was no delay in developmental milestones. The reduced plasma lipid levels change the structural integrity of retina and RBCs causing retinopathy as well as abnormally shaped red cells, low ESR. Peripheral blood smear was suggestive of acanthosis in the present case. Coagulation abnormalities are due to reduced levels of coagulation factors. Cardiac muscle myopathy/ neuropathy or lipofuscin deposition affect these organs' function. Gastrointestinal tracts as well as neurological malignancies have been described in ABL patients.[10]

Patients with ABL are usually asymptomatic at birth but they develop gastrointestinal symptoms during infancy. In infancy, the diagnosis is usually made secondary to failure to thrive. Gastrointestinal manifestations of ABL includes diarrhoea, steatorrhea, chronic fat and fat-soluble vitamins malabsorption resulting in failure to thrive and deficiency of fat-soluble vitamins. These symptoms are worsened with diet rich in fat and improve with avoidance of fatty meals.[2,4,10] Our case also presented with gastrointestinal symptoms at the age of 2-3 months.

Oral diseases can have a direct and devastating impact on the general health and quality of life. Individuals with certainsystemic health problems or conditions such as compromised immunity may be especially vulnerable to the effects of oral diseases. Such patients may express a greater level of anxiety about dental care than those without a disability, which may adversely impact the frequency of dental visits and oral health. These patients require additional considerations for behaviour guidance including the patient's development, education level, cognitive ability, cooperation in medical settings, triggers for uncooperative behaviour, soothing strategies, adherence to schedule or routine, current therapies, and other beneficial accommodations as these can complicate the delivery of care.[11] Consequently, we made an attempt to restore the dental health of the patient with minimal discomfort and anxiety using non-pharmacological behaviour management techniques.

CONCLUSION

Early diagnosis and management of ABL patients is important in preventing devastating long-term serious clinical sequelae. Patients with ABL may not have any distinct oral presentation but providing both primary and comprehensive preventive and therapeutic oral health care to such individuals is an integral part of the specialty of pediatric dentistry.

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