Dental findings in a child with glycogen storage disease type IA

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Glycogen storage disease type I, also known von Gierke's disease, is a rare, severe autosomal recessive disorder due to a defect in liver, kidney, and intestinal mucosa. The existence of delayed development of the dentition, increased incidence of dental caries, taurodontism, and prolonged bleeding following dental procedures should lead clinicians to consider type I glycogen storage disease. A 10-year-old boy with glycogen storage disease type I whose condition was first diagnosed when he was 4 years of age, was referred to the clinic for multiple caries and evaluation of delayed tooth eruption. On physical examination, the patient was cooperative, with short stature, protuberant abdomen, and growth retardation. Laboratory findings indicated that blood levels of pyruvate, triglycerate, uric acid, and cholesterol were elevated. Intraorally delayed mixed dentition was evident, and approximal caries were found in teeth 55, 54, 52, 51, 61, 62, 65, 74, 84, and 85. The most significant radiographic finding was consistent with taurodontism of the molar teeth. Lateral and posteroanterior cephalometric films showed that dimensions of the craniofacial complex were strongly reduced. Evaluation of the patient's dental age was approximately 6 years. *(Quintessence Int 2007;38:9.e36–40)*

Key words: caries, delayed dentition, glycogen storage disease type IA, taurodontism

Glycogen storage disease type I, also known as von Gierke's disease, glucose-6-phosphatase deficiency, hepatorenal glycogen storage disease, or GSD I, is a rare autosomal recessive inborn error of metabolism that results from deficiencies of the microsomal glucose-6- phosphate hdyrolase system. Diminished blood glucose response to glycagon, epinephrine, or galactose infusion is an indicator of glucose-6-phosphatase deficiency. Definitive diagnosis of glycogen storage disease type I is made from a liver biopsy for glycogen determination and enzyme assay directly from the tissues.[1–3] Clinical manifestations of glycogen storage disease type I are characterized by growth retardation,[1,2,4–8] short stature,[8] doll-like face with fat cheeks,[8] and protuberant abdomen that is due to hepatomegaly.[5,7–8] In addition, delayed development of the dentition[8–10] and increased incidence of dental caries are found in some cases.[8–12]

Biochemical findings of the disease include hypoglycemia, lactic acid acidosis, hyperuricemia, and hyperlipidemia. Easy bruising and epitaxis are associated with a prolonged bleeding time as a result of impaired platelet aggregation. Cholesterol and phospholipids are also elevated.[4,7–8]

In 1968, Senior and Loridan[13] first suggested that patients with glycogen storage disease type I might be separated into 2 subtypes. Glycogen storage disease type IA results in a deficiency in glucose-6-phosphate activity, whereas deficiency in microsomal glucose-6 phosphate transport system was found in glycogen storage disease type IB.[2] This case report illustrates the clinical and dentofacial findings in a boy with glycogen storage disease type IA.

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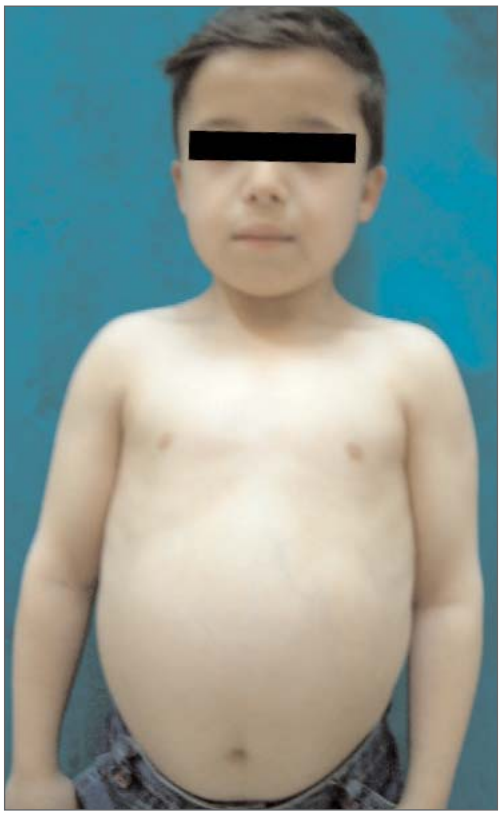

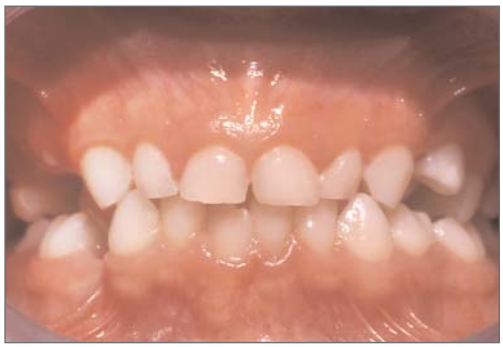

Fig 1 *(left)* Frontal view of patient. Note the protuberant abdomen and short stature.

Fig 2 *(above)* Intraoral view of patient. Note the delayed mixed dentition.

CASE REPORT

A 10-year-old boy with glycogen storage disease type IA whose condition was first diagnosed when he was 4 years of age was referred to the Department of Pedodontics of Ondokuz Mayis University because of multiple caries and evaluation of delayed tooth eruption.

Medical history

The patient was delivered as the second offspring of healthy parents after full-term gestation. At birth, he was 2,500 g in weight and measured 41 cm in height. Consanguineous marriage was present in his family, and the patient's medical history was significant for a number of complications: In 1999, the left kidney was surgically removed under general anesthesia because of renal insufficiency, and the patient had numerous hospitalizations for treatment of hypoglycemia and several instances of spontaneous excessive nosebleeds.

The patient was admitted to the Department of Paediatrics of Ondokuz Mayis University in July 2001. On physical examination, the patient was cooperative, with short stature, protuberant abdomen, and growth retardation (Fig 1). The values for his height and weight were 108 cm and 29 kg, respectively. All vital signs, cardiac ultrasound examination, ECG, and EMG were normal. Abdomen ultrasound examination confirmed hepatomegaly.

Laboratory findings indicated that blood levels of glucose were 51 mg/dL, triglycerate 471 mg/dL, uric acid 9.4 mg/dL, cholesterol 309 mg/dL, and platelets 591,000 mm^3.

Dental and craniofacial findings

Intraorally delayed mixed dentition was present; Teeth 64 and 81 were exfoliated spontaneously approximately 1 year previously (Fig 2). All permanent molars erupted at the age of 8 years and were normal in size and shape. Approximal caries were found in teeth 55, 54, 52, 51, 61, 62, 65, 74, 84, and 85.

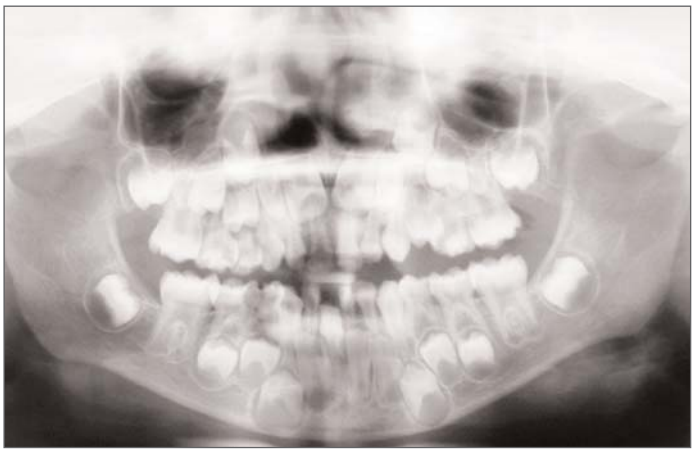

Fig 3 *(above)* Panoramic radiograph showing taurodontism of molar teeth.

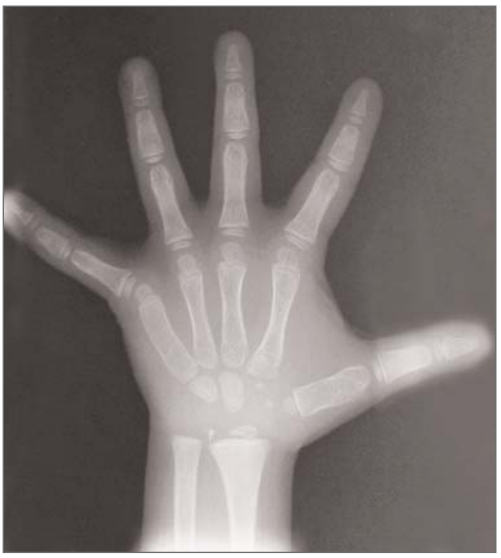

Fig 4 *(right)* Hand and wrist radiograph showing delayed growth and development.

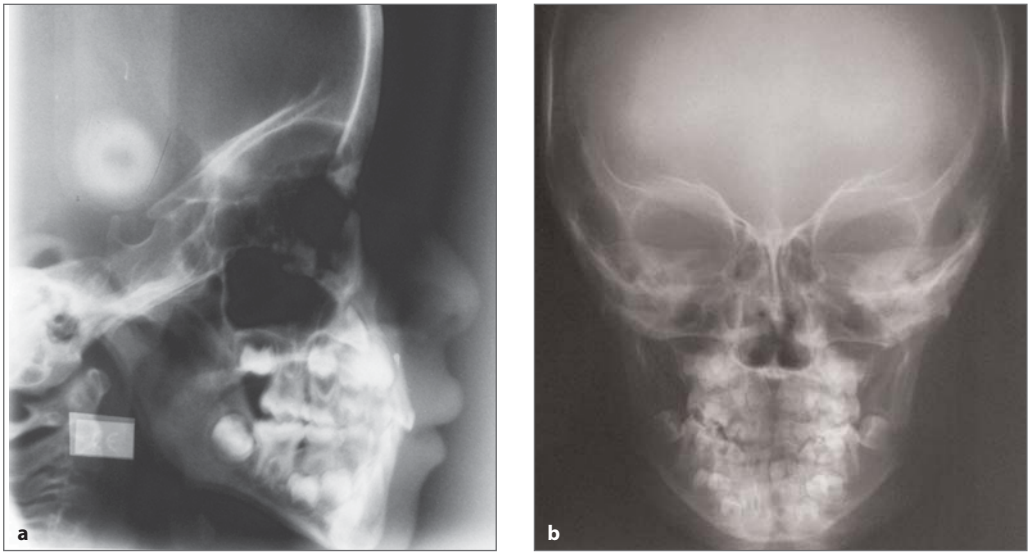

Fig 5 Lateral *(a)* and posteroanterior cephalograms *(b)* show that dimensions of the craniofacial complex are strongly reduced.

Radiographic examinations included orthopantomograph, radiograph of the hand and wrist, standardized lateral cephalogram, and posteroanterior cephalogram (Figs 3 to 5). The most significant radiographic finding was consistent with taurodontism of both primary and permanent molars. The developing tooth germs were normal in number and coronal shape, but their mineralization was delayed. Radiographs of his hands and wrist showed that the carpal bones were delayed in their growth and development. Lateral and posteroanterior cephalograms showed that dimensions of the craniofacial complex were strongly reduced in the child. These findings indicated that the evaluation of the dental age was approximately 6 years.

Treatment

Teeth 52, 51, 61, 62, 65, 74, and 85 were restored with compomer, and teeth 55, 54, and 84 were extracted under local anesthesia because of carious involvement of the pulps after consultation with the Department of Pediatric Hematology. Fissure sealants were applied to all permanent first molars to

Table 1 **Previous case reports of dental findings in a child with glycogen storage diseases (GSD)**

Authors	Type of GSD	Age of patient(s)	Dental findings
Cudzinowski[8]	GSD Ia	13	Dental caries Severe gingival problems
Loevy et al[10]	GSD Ia	8–14	Delayed eruption Dental caries Gingivitis
Katz et al[5]	GSD Ib	3	Dental caries Recurrent neutropenic ulceration
Ralls et al[12]	GSD Ia	29	Gingival bleeding
Bacetti et al[18]	GSD III	4	Deficient craniofacial development Taurodontism
Barrett et al[19]	GSD Ib	22	Rapidly progressive periodontal disease Recurrent mucosal ulceration
Morisaki et al[20]	Fanconi's syndrome + GSDXI	12	Delayed eruption Hypomineralization
Thornhill et al[21]	GSD V	21	Myofascial pain dysfunction syndrome

prevent caries, and fluoride was applied topically. After 3-month follow-up visits over 1 year, no clear increase in dental development was observed, and oral hygiene and restorations were found to be satisfactory.

DISCUSSION

Treatment of glycogen storage disease type 1 patients is aimed at correcting their severe growth retardation by providing exogenous glucose at rates that slightly exceed those of normal hepatic glucose production. This includes frequent carbohydrate feedings during the day and continuous overnight intragastric infusions of glucose or glucose polymers.[4,15] Frequent feeding and a high-carbohydrate diet may lead to development of caries and materia alba accumulation, resulting in enamel decalcification.[5,16] Additional factors, such as chronic metabolic acidosis, may have contributed to the formation of the dental disease.[5] Some researchers suggested using uncooked cornstarch instead of glucose polymers in the diet, as the potential for caries from uncooked cornstarch is less than that of glucose.[17] Kidd et al[11] examined 21 children between 2.7 and 15.5 years of age with hepatic glycogen storage disease and determined that caries were fewer in 2-year-old children than in older children because the younger ones were on the newer diet, which contained uncooked cornstarch. Findings of the present case agree with Kidd et al, as caries were found only in primary teeth, whereas caries-free permanent teeth were present and related to a high-carbohydrate diet until 6 years of age.

Table 1 summarizes previous case reports of dental findings in a child with glycogen storage disease. Delayed dental and craniofacial development is usually seen in patients with glycogen storage diseases. Bacetti et al[18] believe that dental and craniofacial features during the primary dentition phase may be helpful in the early diagnosis of mild forms of the disease and should be regarded in treatment planning for the syndrome. In particular, a strong reduction in the dimension of the craniofacial skeleton agrees with weight and height deficiencies related to the syndrome. Loevy et al[10] believe that delayed dental development, like understature, is secondary to disturbed biochemical findings that result from deficient hepatic and renal glucose-6-phosphatase activity.

Bleeding diathesis is a major consideration in the treatment plan.[12] Primary closure of extraction sites should be avoided to prevent the formation of the hematomata, and

bleeding should be controlled by local measures, including compression and packing with hemostatic agents.[7] The patient experienced several spontaneous excessive nosebleeds. For this reason, local infiltration anesthesia was preferable; mandibular blocks are generally contraindicated because of the possibility of bleeding into the submandibular and subglottic areas into the mediastinum.[22] All extraction sites were packed with Gelfoam and sutured with 3-0 black silk, and adequate hemostasis was achieved.

Bacetti et al[18] found taurodontism of the primary molars in addition to caries in their patient with glycogen storage disease type III. In the present case, the pulp/crown ratio of all molar teeth showed moderate taurodontism, as mentioned by Jorgenson et al.[23] There are no previous case reports that illustrate taurodontism in glycogen storage disease type IA.

Dental findings and treatment management for patients with glycogen storage disease type IA were presented. In conclusion, the pediatrician and the pediatric dentist have to collaborate and gather detailed laboratory, radiographic, and clinical information during the early ages of the affected individuals.

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