原文題目(出處)：	An 18-month-old child with infantile pompe disease: Oral signs. Case Rep Dent 2017, Article ID 5685941
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內文：

1. Introduction

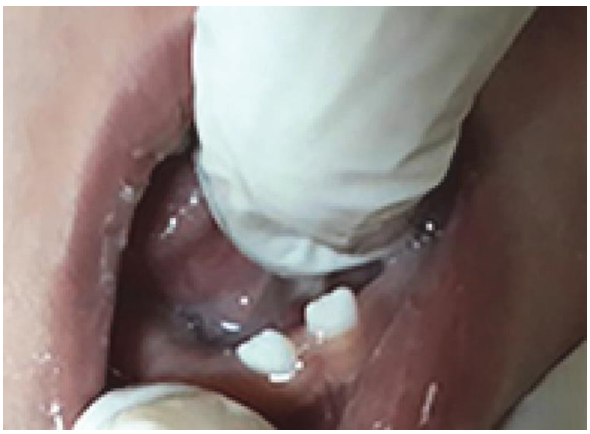
- Pompe disease is an unique glycogen storage disease with a lysosomal metabolism defect and autosomal recessive trait
- This is a relatively rare, fatal disease with a reported incidence of 1 : 14.000–1 : 300.000
- Pompe disease has infantile, juvenile, and adult forms,and the most severe form is the fatal infantile form
- The signs of the disease are progressivemuscle weakness, hypotonia, myopathy, respiratory failure, hepatosplenomegaly, difficulty in feeding, aphonia, weakness in tongue, macroglossia, and inability to control oral secretions
- In the limited number of accessible reports on Pompe disease, orodental findings have rarely been encountered.
- In this case report, we aim to present orodental signs of Pompe disease and contribute to the literature.

2. Case Report

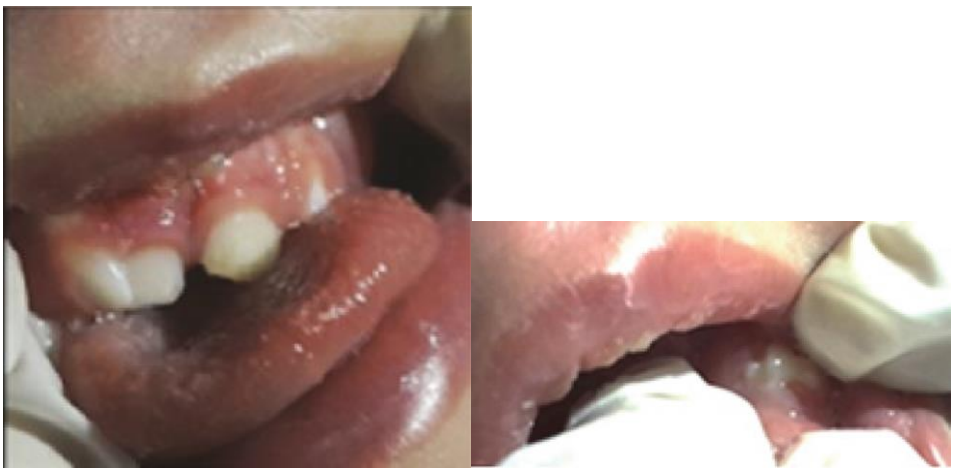
- An 18-month-old male patient with Pompe disease was referred to our clinic due to swelling of his gingiva while he was being treated by enzyme replacement therapy in the intensive care unit of the university hospital.
- a birth weight of 3.270 g
- The other child of the family had died at 16 months due to Pompe disease, and the patient had a 16-year-old healthy brother.
- The medical history of the patient revealed severe muscle hypotonia, respiratory problems, gastric reflux, and a mild cardiomyopathy. His respiration was through a ventilator and he was fed by gastrostomy tube; he was conscious and cooperative but could not speak
- His first dental examination was performed by us when he was hospitalized at the intensive care unit.
- His mouth was found to be open due to macroglossia and respiratory failure, and a nonfluctuant, normal gingiva colored swelling at the right anterior region of the

maxilla was detected

- No color change, focus of bleeding, cystic formation, or infection was encountered at the region. Tooth eruption was noted for 61, 71, and 81 numbered teeth(Figure 1)



- His anamnesis, obtained from his mother, revealed that gingival swelling was also present when 61 numbered tooth erupted.
- His parents were recommended to perform finger massage to the region of 51 numbered tooth. At the examination six months later,51, 52, 62, and 74 numbered teeth had erupted (Figures 2 and 3), and there was fusion between 51 and 52 numbered teeth,and an enamel fracture at the mesial edge of 61 numbered tooth, which his mother was unaware of it, was thought to be secondary to intubation
- 84 numbered tooth was seen to be erupted and it was learned that a swelling at the site of this tooth, similar to the previous one, was present. His parents were recommended to finger massage to this area as well, and no antibiotics or anti-inflammatory drugs were prescribed during this process.The swelling was found to have decreased at the follow-up, one week later.



3. Discussion

- Pompe disease has an autosomal recessive trait, and prenatal diagnosis is generally possible .
- These patients can be examined with the suspicion of tooth agenesis, and a radiography and/or mutational analysis of the genes frequently handled in tooth agenesis could be added to the evaluation for a definitive diagnosis.
- DeGijt et al. [4] reported delayed tooth eruption in the primary molar teeth of a 3-year-old patient who was brought with repeated gum swellings and diagnosed as Pompe disease; however, they reported that the eruption of the permanent teeth of the patient was normal during the follow-up.
- Their histopathological examinations suggested that glycogen accumulation in the fibroblasts might have caused gingival swelling; however,since a significant difference was seen in the glycogen accumulation only in the arterial smooth muscle cells, the etiology of the gingival swelling could not be completely clarified.
- Chronic inflammation, dry mouth, and glycogen accumulation in the fibroblasts have all been considered to play a role in the etiology of swelling

- No report has been encountered in the literature on developmental tooth abnormalities in patients with Pompe disease
- Baccetti et al. reported the presence of taurodontism of the primary dentition and craniofacial disproportion in the patients with glycogen storage disease type III and explained this by the anomalies in the gene content of the X chromosome or less specific abnormalities in chromatinic material development.
- So, the tooth eruption problems, fusion and probable hypodontia, and developmental dental abnormalities that we observed in the case should be included in the signs for Pompe disease.
- Furthermore, rapidly progressive periodontal disease, oral ulcerations, delayed tooth eruption, and gingival inflammation had been reported for glycogen storage disease type I .

- In a study, only a small proportion of the children had caries experience, but most were found to have plaque on both primary and permanent teeth, associated with the inadequate oral hygiene and eating uncooked cornstarch.
- Our patient has not any caries or plaque, related to feeding with gastrostomy tube.

- We consider that the orodental signs of Pompe disease are inadequately emphasized in the limited number of accessible sources in the literature.

- The fact that Pompe disease is not one of the subjects which dentists commonly come across may be due to the fact that patients with Pompe disease are hospitalized in the intensive care units, their life expectancy is short, and the disease is very rarely seen.
- We hope that, by presenting information on the oral signs of Pompe disease with this case report, a source can be provided in order to assist in the diagnosis and treatment of children with this disease and to establish dentistry care for those patients.

題號	題目
1	Pompe's disease 是一種肝醣儲存疾病（glycogen storage disease），是因為缺乏下列何種酵素活性所引 起？ (A) glucose 6-phosphatase (B) lysosomal glucosidase (C) debranching enzyme (D) phosphorylase kinase
答案 (B)	出處：101 年 - 101 年第一次生物化學與臨床生化學
題號	題目
2	Which disease may not cause hypodontia? (A) Down syndrome (B) ectodermal dysplasia (C) cleft lip and palate (D) idiopathic hypoparathyroidism (E) hyperthyroidism
答案 (E)	出處： 1. Hypodontia in patients with Down's syndrome.

	-Department of Orthodontics, School of Dental Medicine, University of Zagreb 2. Dental findings in patients with ectodermal dysplasia -Institut für Kieferorthopädie, Orthodontie und Kinderzahnmedizin Centrum für Zahn-, Mund- und Kieferheilkunde, Charité-Universitätsmedizin Berlin, Assmannshauser Str. 4-6, 14197 Berlin, Germany. thomas-michael.praeger@charite.de 3. The occurrence of hypodontia in patients with cleft lip and/or palate. - Department of Cleft Defects, University Hospital Královské Vinohrady, Prague, Czech Republic. 4. Nature and frequency of dental changes in idiopathic hypoparathyroidism and pseudohypoparathyroidism. - Jensen SB, Illum F, Dupont E