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內文：

一、Abstract

Zellweger syndrome (ZS)

1. rare autosomal recessive disorder(染色體隱性遺傳病)
2. resulting from an impairment in peroxisome(過氧化小體)function.
3. It is characterized by craniofacial dysmorphism(顱顏畸形)and neurological abnormalities(神經性的異常),and involves several systems, which may complicate dental and anesthesia management.
4. Collaboration with the medical team is essential for optimal care of these patients.

二、Introduction

1. also called cerebrohepatorenal syndrome 腦肝腎綜合徵
   an autosomal recessive disorder caused by mutation in any one of several genes involved in peroxisome biogenesis
2. Prevalence：
   (1) 1:100,000 live births
   (2) milder variants and atypical cases : 1:25,000 to 1:50,000.
3. Facial abnormalities：
   (1) high forehead(2)large fontanels(3)hypoplastic supraorbital ridges, (4)epicanthal folds 眼內贅贅皮(5)flat and broad nasal bridge(6)external ear deformities 外耳病變(7)micrognathia(8)high arched palate
4. Neurological abnormalities：severe hypotonia 肌張症, abnormal Moro reflex 擁抱反射, hypo- or a-reflexia, epileptic seizures,and psychomotor retardation 心理運動遲緩.
5. D.D.: Down syndrome, Prader–Willi syndrome & spinal muscular atrophy
6. Clinical data : elevated plasma and tissue levels of very-long-chain fatty acids, phytanic acid 植烷酸, pipecolic acid 哌啶甲酸, and bile salt precursors.Decreased plasmalogen 縮醛磷脂 synthesis is also noted
7. Prognosis : poor
8. Prenatal diagnosis : immunoblotting of peroxisomal β-oxidation enzymes

三、case report

■ Data

1.age：7-year old

2.P.I.：this 7 y/o female patient was brought to Pediatric Dentistry Clinic, Faculty of Dentistry, Khon Kaen University because of her dental caries and poor oral hygiene.

3. Family history(hereditary diseases)：NP

4.Her parents and her 6 y/o brother：normal stature

■ Medical and dental history
  Medical history :
  Multiple congenital anomalies presented at birth
  neonatal period : treated with O2 supplementation for 1 month(asphyxia窒息)

sensorineural hearing loss(感覺神經性聽障), severe psychomotor retardation(心理動作遲鈍)delayed development, chronic bronchiectasis(慢性支氣管擴張症).

7 months old : diagnosis of ZS

11 months old : hypothyroidism

2 years old : Seizures began , frequent hospital admissions

4 years old : gastrostomy 胃造口術 and tracheostomy 氣管造口術

Dental history : OHI (6-yr old)

■ Examination

1. physical examination：

typical dysmorphic facial features(顏面畸形). Fig-1

Dolichocephaly(頭形長), prominent forehead, hypertelorism(眼距過寬), shallow supraorbital ridges, epicanthus(內此贅皮), flat & broad nasal bridge, anteverted nostrils(鼻孔前傾),long philtrum(人中過長), micrognathia(小頜畸形)…etc.

2. Intraoral examination：

(1) Heavy deposition of plaque&calculus,esp on mandibular teeth

(2) Ankyloglossia and generalized spacing of mandibular teeth.

(3) Vault: Lateral part(low in height) middle part (dome-like shape).

(4) Maxillary incisors were crowded

(5) The primary canines and first molars had not fully erupted.

(6) Carious lesions : palatal surfaces of both maxillary central incisors and occlusal surfaces of both maxillary first molars.

(7) Class III malocclusion with anterior open bite

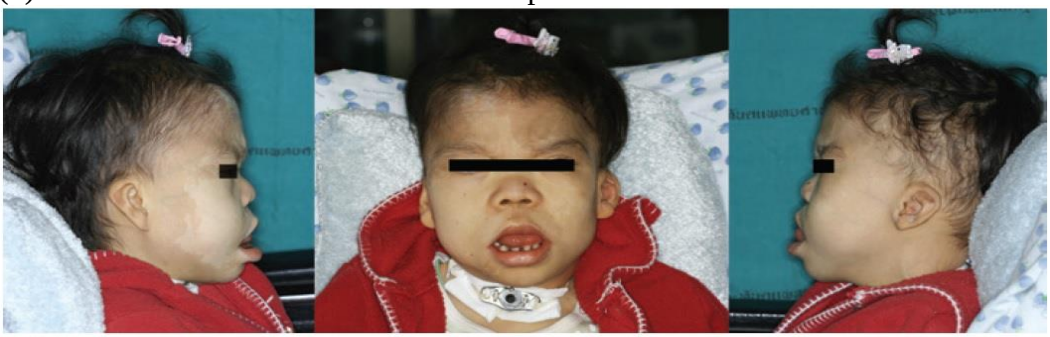

Figure 1. Typical craniofacial dysmorphic features seen in the patient.

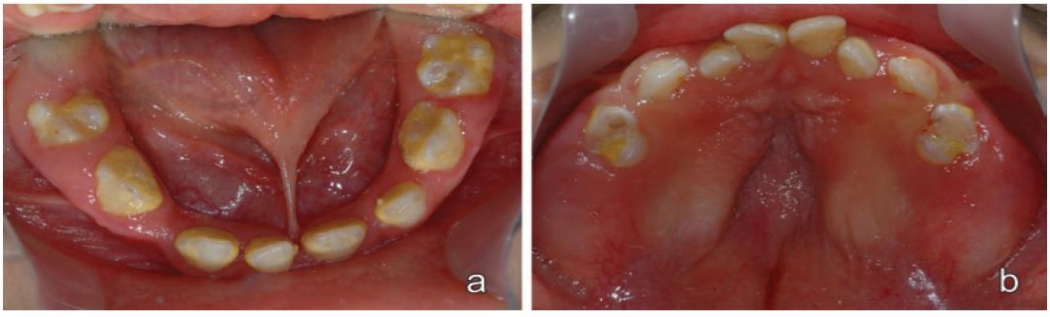

Figure 2. Intraoral appearance showing heavy plaque and calculus deposits, delayed eruption, (a) ankyloglossia, spacing and missing mandibular incisors, (b) crowding maxillary incisors and distinctive palatal morphology.

3. Radiograph examination：

delayed dental development and microdontia of mand. 2nd permanent molars

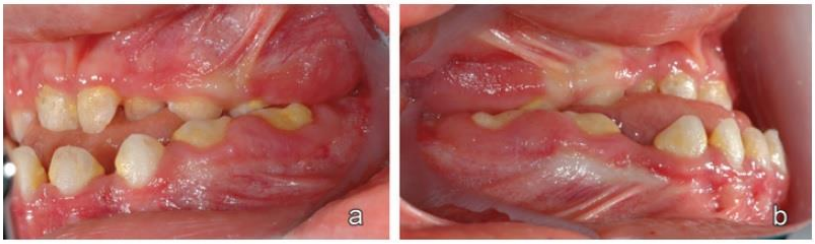

Figure 3. Right (a) and left (b) occlusion displaying Class III malocclusion with anterior open bite.

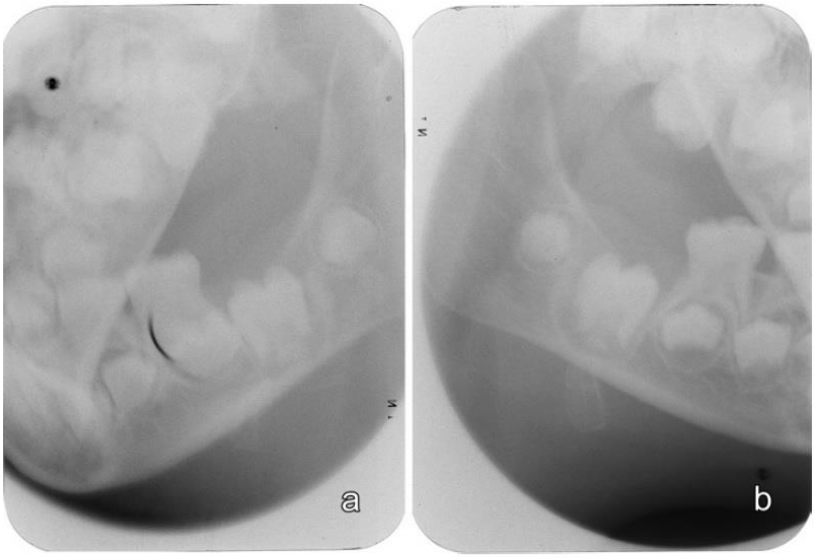

Figure 4. Right (a) and left (b) lateral jaw radiographs showing delayed dental development and microdontia of second permanent molars.

■ Anesthetic and dental management

Dental procedures consisted of radiographs, full mouth scaling and prophylaxis, placement of composite restorations on maxillary central incisors,preventive resin restorations on maxillary first molars and sealant on mand.1st molars, and full mouth fluoride varnish application.

Oral hygiene instruction to the mother was reinforce.

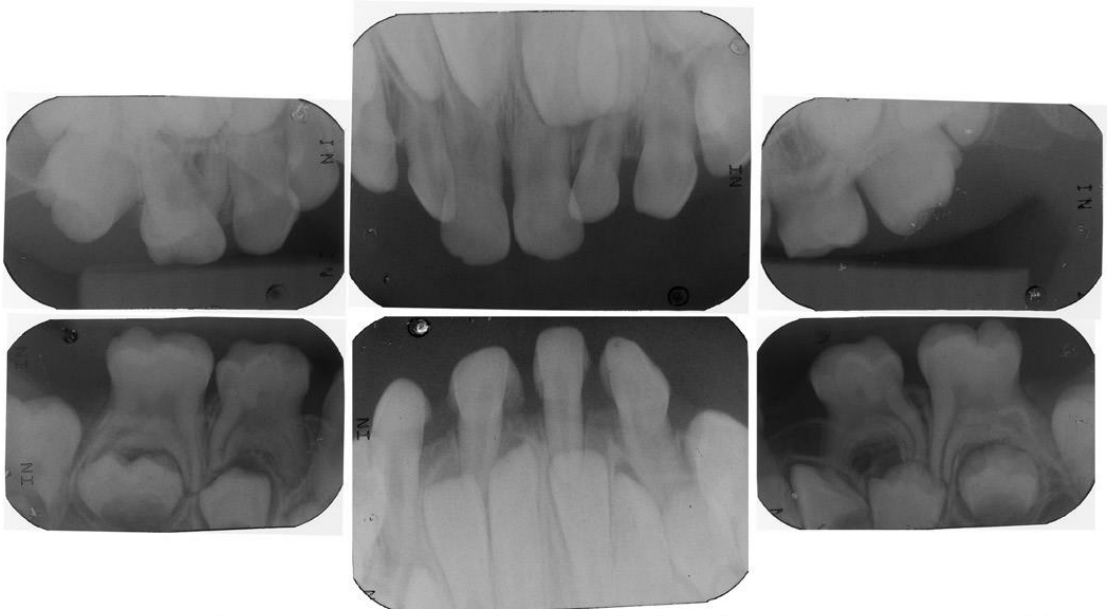

Figure 5. crowding max. incisors, heavy calculus deposits, missing mandibular incisors and delayed molar eruption.

四、discussion

1. most cases of ZS :
   (1) lethal in early childhood(1~3 歲)
   (2) several reports of survival into late childhood (7~9 歲)and adulthood
2. This case :
   (1) survived longer than usual
   (2) totally dependent and has had multiple hospital admissions
   (pneumonia 肺炎, diarrhea, secretion obstruction, infection & failure to thrive)
   (3) distinctive palatal morphology (differed from high arched palate)
   (4) With clinodactyly 第五手指彎斜向內(differed from Camptodactyly 屈曲指)
   (5) congenitally missing teeth, malocclusion, and delayed dental development
   (6) tooth eruption : average dental age of a 2-year-old child
   calcification of developing permanent teeth : dental age of a 5-year-old child
   (7)present with respiratory insufficiency
3. Sedative premedication : not recommended (risk of airway obstruction)
4. The patient was fed via gastric tube:
   (1)heavy plaque and calculus deposition : greatly diminished swallowing reflex can result in salivary stagnation(唾液停止分泌)
   (2) more aspirationpneumonia 吸引性肺炎 - associated microorganism in saliva
5. Rapid formation of calculus : requires frequent dental follow-up
6. Home-care regimen : regular flossing and brushing with fluoride toothpaste

五、conclusion

This case report shows that it is possible for children with ZS to survive at least into later childhood(9yr~12yr).

The comprehensive dental care for these children can be difficult because they possess several medical and dental problems.

Therefore, the collaboration with medical team is essential.

題號	題目
1	下列何者不是 Down syndrome 的臨床表徵? (A) Protruding tongue (B) Microglossia (C) Flat midface (D) Delayed eruption of permanent teeth
答案(B)	出處：Oral and maxillofacial pathology 3rd edition , Neville, et al
題號	題目
2	關於 Prader–Willi syndrome 的敘述何者錯誤？ (A) 為 13 號染色體上一段編碼小核內核糖核蛋白(SNRPN)基因缺陷 (B) 常出現手、面部畸形(face malformation) (C) 常出現明顯肌張力低下(muscle hypotone) (D) 在嬰幼兒期會出現斜視(Strabismus)、脊椎側彎(Scoliosis)
答案(A)	出處：