

## Case Report

## Kocher Debre Semelaigne syndrome and associated orofacial aspects: report of a case

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**Abstract:** Myopathy and muscular pseudohypertrophy associated with severe congenital hypothyroidism has been described as Kocher Debre Semelaigne syndrome, which is a rare disorder with only a few previously published reports. However, none of these reports have described the orofacial aspects of this syndrome. Here we report one such case, which may provide valuable information to specialists in oral healthcare regarding the associated orofacial aspects. (J Oral Sci 53, 129-132, 2011)

**Keywords:** Kocher Debre Semelaigne syndrome; hypothyroidism; orofacial aspects.

### Introduction

Myopathy and muscular pseudohypertrophy associated with severe congenital hypothyroidism has been described as Kocher Debre Semelaigne (KDS) syndrome, also known as cretinism-muscular hypertrophy (1). This is a rare disorder usually seen in cases of congenital hypothyroidism, but it may also be associated with acquired hypothyroidism. Only a few cases of KDS syndrome have been reported previously. It occurs more commonly in males, and has been reported in children who are the products of consanguineous marriages. Here we present one such case to emphasize the orofacial aspects of this rare syndrome.

### Case Report

A 13-year-old girl with severe congenital hypothyroidism

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was referred to our department from the Department of Pediatrics for evaluation of dental caries. The patient was mentally retarded and her developmental milestones had been delayed. She presented with features typical of hypothyroidism, including reduced appetite, chronic constipation with hard stools, somnolence, cold intolerance, delayed response to verbal commands and visual stimuli, and poor fine motor skills. The patient started speaking at 7 years of age and could speak only a few words.

The patient was the third of the four children in the family (Fig. 1). She had been the product of a full-term normal delivery, and her other siblings and parents (non-consanguineous marriage) were in good general health. General examination revealed that the patient had a short stature (101 cm) (Fig. 2) and subnormal weight for age (16.5 kg), with a BMI of 16.4. Other features included a short neck, umbilical hernia, prominent calf muscles with proximal muscle weakness in the lower limbs, coarse skin (Fig. 3), incoherent speech and a hoarse voice.

Extraoral examination (Fig. 4) revealed that the patient had an acceptable facial symmetry, a dull look on the face, a sparse hairline at the forehead, rough hair texture, a broad forehead with thick skin, coarse facial features, a depressed nasal bridge, hypertelorism, nystagmus, puffed

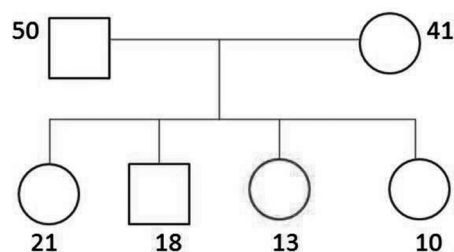


Fig. 1 Figure showing the patient's pedigree chart.

lips, protruding tongue, a poorly developed mentolabial sulcus, lower face deficiency, mandibular hypoplasia and retrognathism. A TMJ examination showed nothing remarkable and there was no lymphadenopathy. However, firmness suggestive of a 1 × 1 cm mass was felt in the submental region on palpation.

Intraoral examination showed that the patient’s oral hygiene status was poor. Mixed dentition was seen with permanent incisors and the first molar, and deciduous

canines and molars in each quadrant. Deep dentinal carious lesions with pulpal involvement were noted in relation to 54, 55, 16, 63, 64, 65, 26, 73, 74, 75, 36, 83, 84 and 46. However, none of these teeth were tender on percussion. Root stumps were seen in relation to 75, 85. The palate was highly arched (Fig. 5). The molar relation was flush terminal and anterior open bite was evident (Fig. 6). None



Fig. 2 Figure showing the short stature of the patient.



Fig. 4 Figure showing the abnormal extraoral features.

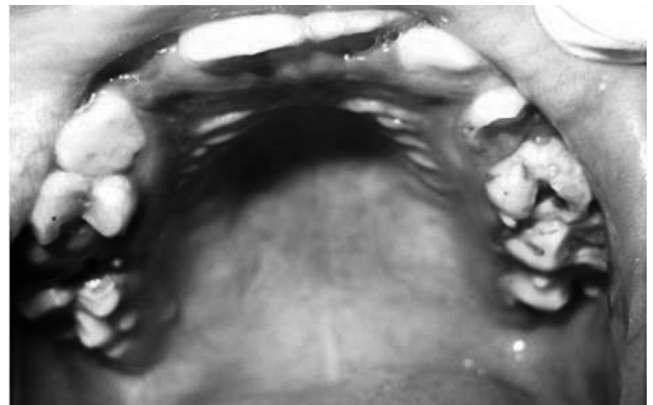


Fig. 5 Figure showing the high arched palate.

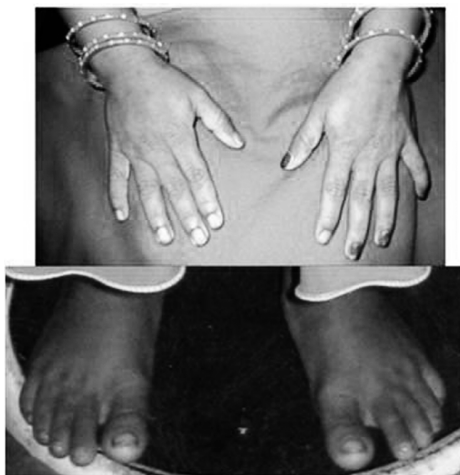


Fig. 3 Figure showing the characteristic coarse skin.



Fig. 6 Intraoral examination revealing anterior open bite.

of the teeth were mobile or tender on percussion. Mild inflammation and melanin pigmentation of the marginal gingiva was seen.

Based on the above features, a clinical diagnosis of skeletal (class 2) and dental malocclusion secondary to hypothyroidism was made. The investigations performed on the patient included a routine hemogram, determination of T3, T4, TSH, and creatine kinase levels, electromyography, and nuclear medicine thyroid scan. The patient performed poorly in the Binet Kamat and Seguin Form tests, which indicated subnormal intelligence. Vineland social maturity scale indicated a social age of 2 years and 5 months, with poor adaptive and social functioning.

The hemogram revealed that the hemoglobin level was low at 8.1 gm/dl. A peripheral smear showed normocytic, normochromic red blood cells, 44% neutrophils, 37.6% lymphocytes, 10% monocytes, 7.8% eosinophils and 0.6% basophils. RBC count: 2.63 million/ $\mu$ l, hematocrit: 24.11%, MCV: 91.5  $\mu$ m<sup>2</sup>, MCH: 30.8 pg, MCHC: 33.6%, RDW: 13.1%, WBC count: 6,800/ $\mu$ l, platelet count: 276,000/ $\mu$ l. The levels of T3 and T4 were significantly depressed at 0.2 ng/ml and 0.9  $\mu$ g/dl, and that of TSH was markedly elevated at 100 IU/ml. Creatine kinase activity was also elevated at 440 units/L. The findings of electromyography were suggestive of myopathy. In the nuclear medicine thyroid scan, static images were obtained 20 min after i.v. injection of Tc 99m pertechnetate, and anterior oblique views were taken. The scintigraphic features were suggestive of poorly functional ectopic thyroid tissue in the submental region, where a small focus of radiotracer concentration was seen (Fig. 7). There was no evidence of functional thyroid tissue over the thyroid bed. Ophthalmology consultation was sought for evaluation of nystagmus and delayed response to visual stimuli, and this attributed the nystagmus to a central cause.

On the basis of myopathy and muscular hypertrophy associated with congenital hypothyroidism, a diagnosis of Kocher Debre Semelaigne syndrome with congenital nystagmus was made.

Investigations planned and performed at our department included panoramic and lateral skull radiographs. The panoramic radiograph demonstrated unerupted permanent canine, premolars and a second molar in each quadrant. A radiopacity was seen in the pulp chamber of 75, which was suggestive of a pulp stone or enamel pearl. Radiographic evidence of deep dental caries was seen in relation to almost all of the posterior teeth (deep dentinal carious lesions were confirmed radiographically in all the affected teeth) (Fig. 8). Lateral skull radiography confirmed a class 2 skeletal profile (with an open bite, high mandibular plane angle, and an ANB angle of approximately 5°) and

reduced dimensions of the facial complex (Fig. 9).

The patient was started on Eltroxin 100  $\mu$ g once daily,

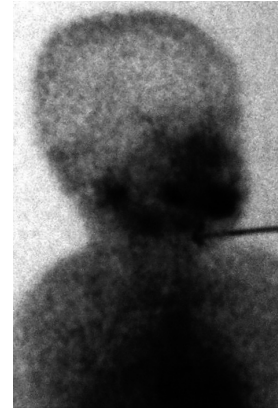


Fig. 7 Figure showing radiotracer concentration in the submental region due to ectopic thyroid tissue.



Fig. 8 Panoramic radiograph showing unerupted permanent canines, premolars and second molars.

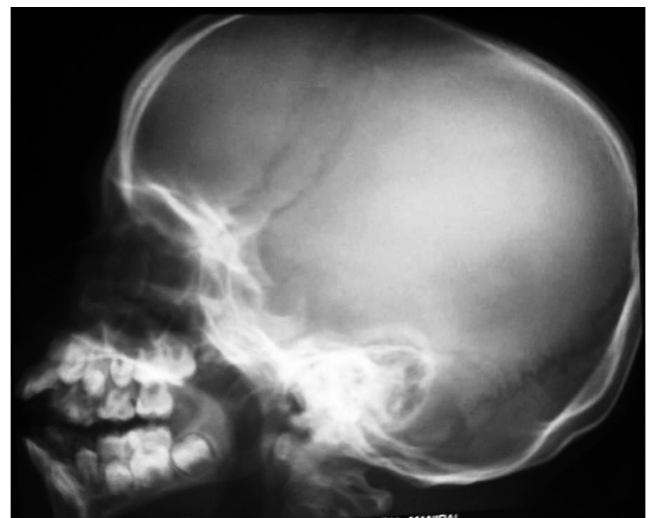


Fig. 9 Lateral skull radiograph showing reduction in the dimensions of the facial complex.

and a comprehensive, multidisciplinary treatment plan was drawn up, consisting of preventive measures such as diet counseling, oral prophylaxis and topical fluoride application, pediatric endodontic procedures and interceptive orthodontic procedures.

### Discussion

In 1898, Kocher described a case of hypothyroidism with muscular hypertrophy, and in 1935, Debre and Semelaigne described the relationship between the two (KDS), characterized by myopathy of hypothyroidism and limb muscle pseudohypertrophy in infancy or childhood. KDS is a rare disorder usually seen in congenital hypothyroidism, but may also be associated with acquired hypothyroidism. It is also known as cretinism-muscular hypertrophy, hypothyroid myopathy, hypothyroidism-large muscle syndrome, hypothyrotic muscular hypertrophy in children, myopathy-mixedema syndrome, or mixedema-muscular hypertrophy syndrome (1). To our knowledge, there have been only a few reported cases of KDS syndrome, and most of the reports have been in French. The usual age at presentation is between 18 months and 10 years, but the condition has also been diagnosed in infants and neonates. The pseudohypertrophy involves the muscles of the extremities, limb girdle, trunk, hands and feet, but it is more prominent in the muscles of the limbs. This condition is seen more commonly in males, and has been reported in children with consanguineous parents; an autosomal recessive inheritance has been suggested (2). However, the present patient was a female and her parents had a non-

consanguineous marriage, thus making this case rarer. Hypertrophy in this condition is most common in the muscles of the calf, thigh and back.

Severe hypothyroidism, such as that in the present case, can have deleterious effects on tooth development and eruption, and lead to prolonged retention of the primary dentition, subnormal growth of the maxilla and mandible with a marked reduction in the dimensions of the facial complex (3), and a lack of coordination between mandibular growth and dental development. However, the dental structures can still respond to the effects of L-thyroxine at a relatively late age, with exfoliation of the primary dentition and eruption of the secondary dentition (4).

### References

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