

## HEARING IN KNIEST SYNDROME

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### ABSTRACT

Kniest syndrome is a type of dwarfism that is characterized by unusually short arms and legs, a round face with hollow or depressed areas, swelling and stiffness of the joints and a stiff drawing up (contractures) of the fingers. Cleft palate, abnormal curvature of spine (scoliosis), and vision and/or hearing problems may also occur. Intellect is usually normal in people with this syndrome.

In this case report we present the audiologic findings of an eight-year-old boy with Kniest syndrome.

**Key Words:** Kniest syndrome, Hearing loss, Audiological evaluation

### INTRODUCTION

The first form of generalized spodyloepimetaphyseal bone dysplasia with disproportionate dwarfism was described by Kniest in 1952 (1,2).

Kniest syndrome is a rare, genetic osteochondrodysplasia. 200 cases are currently known world wide. The disease is autosomal dominant, and due to defects in the secretion of collagen type II

associated with mutations in the COL2A1 gene. Gene map locus of the syndrome is 12q13.11-q13.2 (3).

This disorder is also termed Swiss Cheese Cartilage Syndrome, Kniest Dysplasia or Metatropic Dysplasia II.

The symptoms consist of peculiar faces with midfacial hypoplasia, saddle nose, and occasional shallow orbits with protruding eyes; round flattened face with short neck, short trunk with dorsal kyphosis, lumbar lordosis, and less frequently thoracic scoliosis in the later course of the disease; short and broad thorax with sternal protrusion; short arms and legs with prominent joints and restricted joint mobility; cleft palate; mixed, conductive or sensori-neural hearing loss (1,2,4) myopia; retinal detachment, and club feet. Some manifestations such as short stature, prominent knees, cleft palate and/or club feet may be present at birth, but the full expression usually occurs by the age of 3 years. Intelligence is usually normal.

### CASE REPORT

Our patient, an eight-year-old boy was referred to the Marmara University Sub-department of Audiology for evaluation of his hearing (Fig 1).

He had short-trunk, short-limbs and cleft palate at birth. At the age of 17 months old he was diagnosed with the Kniest syndrome in another institution where he had undergone a genetic investigation.

Besides his short-limbs and cleft palate, almost all his joints: the elbows, wrists, knees and ankles were enlarged (Figs. 2-5). He had thoracic scoliosis, chest deformity and severe myopia. His height was less than P10 (10th percentile) and he had normal intelligence.

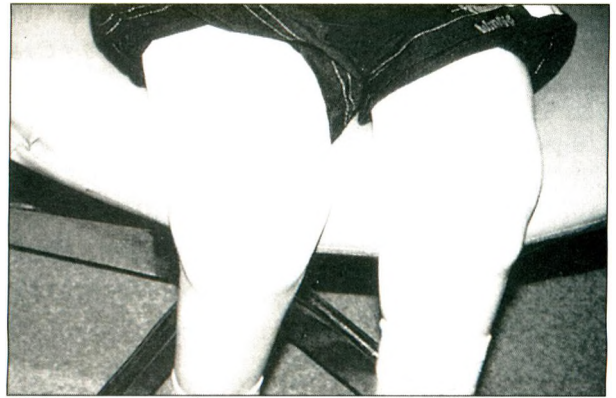
His cleft palate was repaired at the age of 2. He did not walk until 3 years of age. He had an operation on both of his knees when he was 4. He had a waddling gait.

Audiometry indicated a symmetric bilateral mixed

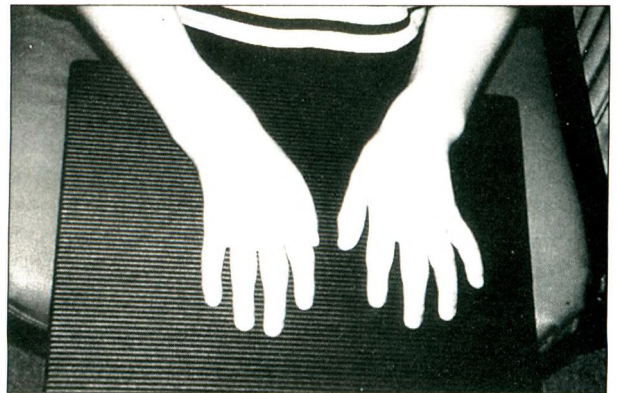


**Fig.2:**

Lumbar lordosis develops within first few years of life. Joints become enlarged and stiff.



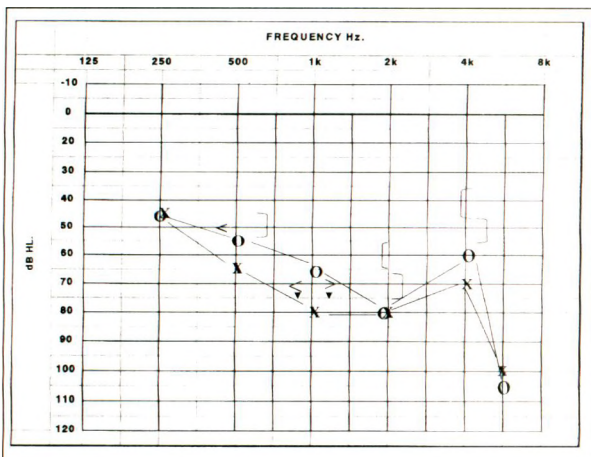
**Fig.3:** Flexion and extension of enlarged knees are reduced



**Fig.4:** Metacarpophalangeal joints are enlarged.



**Fig.5:** The feet are inverted and ankle joints are enlarged.



**Fig. 1:** Hearing thresholds for air conduction right and left ear are shown with O and X. Unmasked bone conduction thresholds are shown with < for right ear and > for left ear. Masked bone conduction thresholds are shown with [ for right ear and with ] for left ear respectively.

type of hearing loss. His pure tone average (PTA) in the right ear was 67 dBHL and 75 dBHL in the left ear. The speech reception thresholds were in agreement with PTA averages. The discrimination score on the right ear was 60% and on the left ear 76%. Tympanometric findings were normal on both ears (type A). Contralateral and ipsilateral

stapedial reflexes could not be elicited in either ear. Transient evoked otoacoustic emissions (TEOAE) were also absent in both ears.

## DISCUSSION

Kniest syndrome is recognizable at a very early age, usually diagnosed at birth by the shortness of the limbs or increase in size of the joints. Myopia, deafness and cleft palate is frequently associated with this syndrome (5-7).

Hearing loss is present in 50% of the patients (4). Conductive, sensori-neural or mixed type hearing losses are frequent findings. (1,2,4) Hearing loss may develop before puberty. Recurrent otitis media and respiratory infections are common (1). Siggers et al. reported 8 cases. Two were identical twins; the other cases were sporadic. Cleft palate was present in 5, deafness in 6, retinal detachment in three (8). Barona et al. described a case in whom they thought the hearing loss was conductive due to the coexistence of a cleft palate. But afterwards when the girl was 12 years old they observed the cochlear component (4). MacDermot et al. presented a family with short stature, femoral epiphyseal dysplasia, mild vertebral changes, and sensori-neural deafness inherited as an autosomal dominant trait (9). Beighton and Goldberg reported an Africaner kindred who had dominantly inherited skeletal dysplasia, blindness and deafness syndrome. The child had reduced stature and a round, flattened face with conductive deafness (10). Tscherninkov and Alexandrov described three women with dwarfism and skeletal defects characteristic for Kniest dysplasia. In two of the patients there were only skeletal defects. In the third patient, they were combined with other symptoms pertaining to the tendons, the sclera, eye lenses, the retinal vessels and loss of hearing (11).

In this report we present a male child with mixed type of hearing loss moderate degree in the right ear, severe degree in the left ear. His hearing deficit on both ears was not noticed until he was examined in our audiology clinic at the age of 8.

The literature contains almost no well-documented reports of longitudinally assessed

hearing. Since Kniest syndrome can be diagnosed at the newborn period, hearing should be tested at this stage and the patient should have periodic hearing measurements.

We want to stress the importance of evaluation of hearing in these patients at the newborn period. Assessments should be made at least at 6 monthly intervals or when parents suspect signs of hearing loss, to find out when hearing impairment develops if it is not present congenitally.

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