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LARSEN'S SYNDROME

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A girl with the characteristic abnormalities of Larsen's syndrome is presented. A soft flabby consistence of the cartilaginous skeleton of the larynx and trachea was thought to be the cause of attacks of respiratory failure which suddenly caused her death at the age of 9 months. Microscopy revealed a considerably reduced number of elastic fibres in the larynx, trachea and bronchi. Closed reduction of the knee dislocation by skin traction seemed to be successful.

Key words: congenital malformations; knee dislocation

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The first description of the disorder later known as Larsen's syndrome was given by Kaijser (1935). He reported a case with multiple congenital malformations including knee dislocation and facial deformities. There was also a family history of congenital deformities of the musculo-skeletal system. Larsen et al. (1950) reported six unrelated patients with multiple joint dislocations, depressed nasal bridge and wide-spaced eyes. The joints most often affected were the knees, elbows and hips. Abnormalities of the foot, hand and spine, and cleft palate were also found.

Since this publication, some reports have appeared describing cases consistent with Larsen's syndrome (Curtis & Fisher 1970, Steel & Kohl 1972, Micheli et al. 1976, Oki et al. 1975). New findings have also been added to the clinical and pathologic-anatomic entity. Latta et al. (1971) reported malformation of the larynx giving respiratory distress. Most authors have found a familial occurrence of the syndrome, and Habermann et al. (1976) reported four cases of Larsen's syndrome in three generations of one family and suggested a dominant mode of inheritance.

The purpose of the present paper is to report a case of Larsen's syndrome in which the patient suffered from attacks of respiratory failure. One such attack resulted in her sudden death at 9 months of age. In this patient closed reduction of the knee dislocation was attempted using skin traction.

CASE REPORT

The patient was a girl, 3 months of age when seen for the first time, at Sophies Minde Orthopaedic Hospital, because of congenital dislocation of her right knee. She was born at full-term to a 33-year-old woman (gravida 4, para 2). Her father and an older sister both had an unusual shape of the face with depressed nasal bridge and hypertelorism. The father also had syndactylism of toes of both feet, and he had had a plastic operation for the flattened nose. The sister had short first metatarsal bones.

Shortly after birth it was noticed that the baby had a dislocation of her right knee. Her face had a characteristic appearance with depressed nasal bridge and hypertelorism (Figure 1). The hard palate was high-arched. Calcaneo-valgus deformity and short first metatarsal bones of both feet were found (Figure 2). Examination of the hips revealed a positive dislocation test on the left side. She also

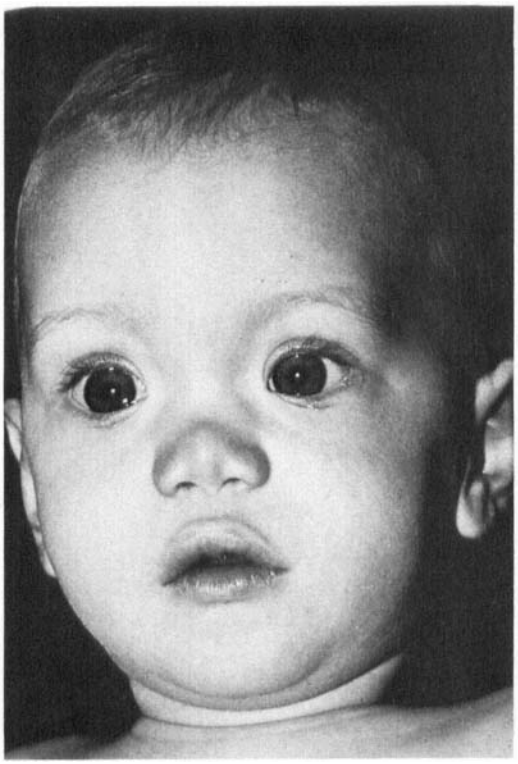

Figure 1. Depressed nasal bridge and ocular hypertelorism in a 6-month-old girl with Larsen's syndrome.

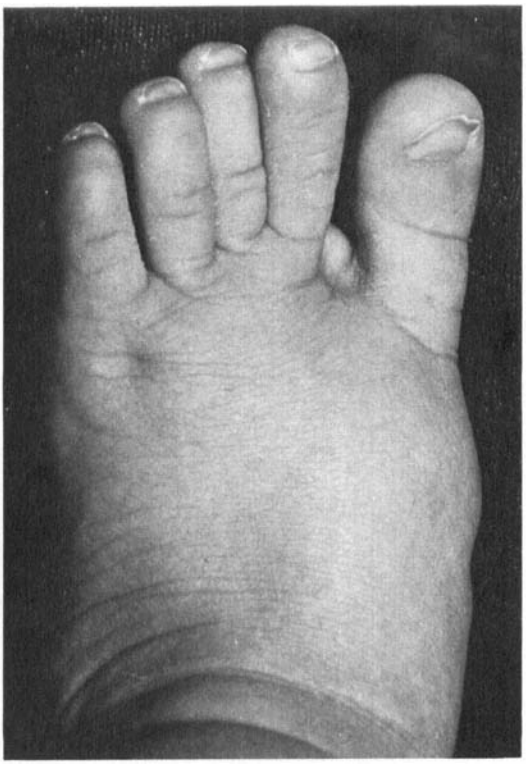

Figure 2. Short first metatarsal bone in Larsen's syndrome.

had a pectus excavatum deformity. Closed reduction of the dislocated knee was attempted but failed. The hip dislocation was treated by a Frejka pillow and the foot deformity by corrective casts.

A few days after birth, attacks of respiratory distress with cyanosis supervened. The attacks were apt to occur during meals, and the patient had to be fed through a stomach tube for some weeks. Direct laryngoscopy showed a soft, easily collapsing epiglottis and hypoplasia of the arytenoid cartilage. During the first 6 months of life the attacks became more infrequent, and when orthopaedic treatment of the knee dislocation started, the respiratory failure was considered cured.

Radiographs of the entire skeleton revealed an anterior dislocation of the right tibia on the femur, a slight dysplasia of the left acetabulum, an extra ossification centre of os calcis and a short first metatarsal bone of both feet. A general muscular hypotony was found. Laboratory findings, including chemical studies of serum and urine and electrocardiograms were normal as was muscle biopsy.

Treatment was started with skin traction, the knee in semi-flexion and countertraction applied on the distal thigh. After 3 weeks of traction radiographs showed that the dislocation was almost reduced to a normal position (Figure 3). However, sudden death supervened at this time due to an attack of respiratory failure.

Necropsy showed a soft, flabby consistence of the epiglottis and arytenoid cartilage. Trachea and larynx were partially collapsed. No signs of aspirated food or cardiac malformation were found. Microscopy revealed a markedly reduced number of elastic fibres in the larynx, trachea and bronchi. Signs of an interstitial pneumonia were also found.

The right patella was connected to the femoral condyles by fibrous tissue occupying the entire suprapatellar pouch. Splitting of the infrapatellar tendon and the anterior part of the capsule led to reduction in the flexed position.

DISCUSSION

A girl having physical findings consistent with Larsen's syndrome is presented. Her father

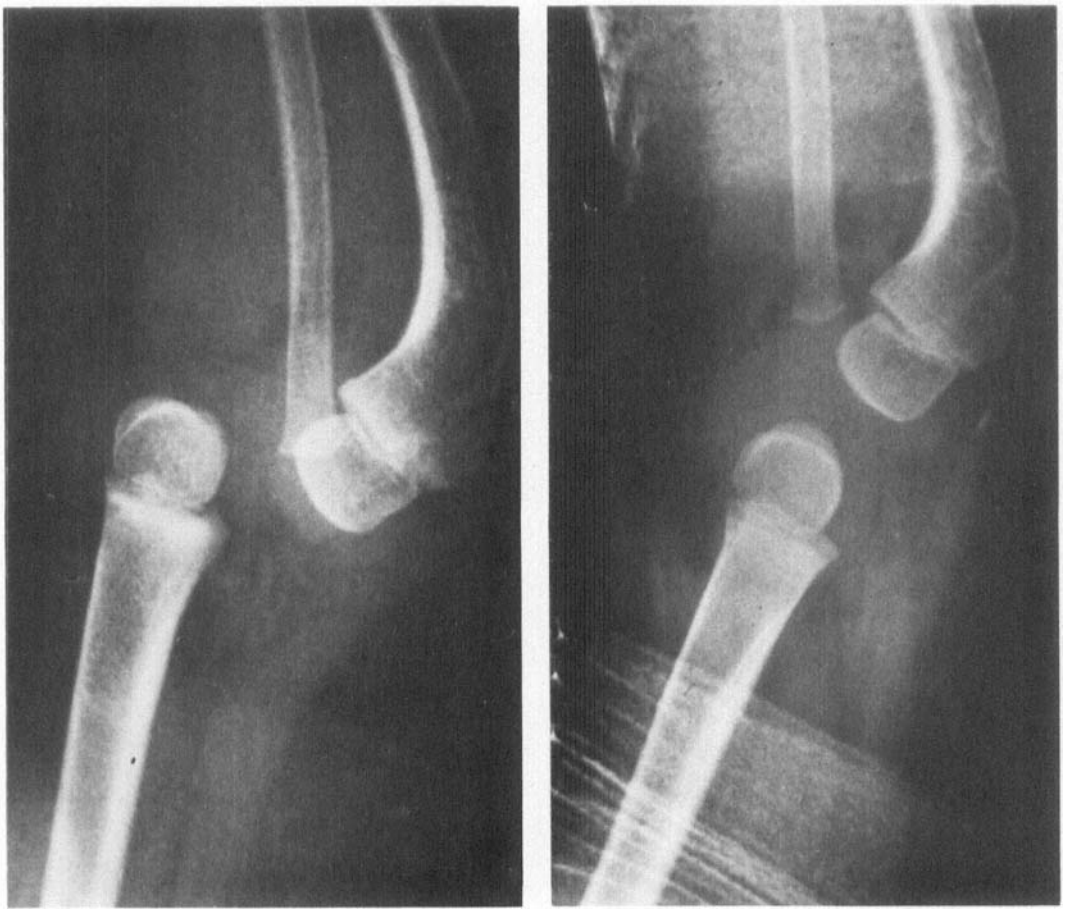

Figure 3. Anterior dislocation of the tibia (a). The knee almost reduced in skin traction (b).

and sister also had some deformities characteristic of this syndrome, and her mother had had two abortions. The family history could point to a dominant mode of inheritance with a wide variety in the penetrance of the disorder. Our case had a distinct maldevelopment of the cartilaginous skeleton of the larynx and trachea, verified at autopsy. Histological examinations revealed reduction of elastic fibres in these structures. This finding has not previously been reported in Larsen's syndrome.

At present Larsen's syndrome is a rather complex and not well defined entity. In Table 1 all abnormalities ever described in cases supposed to have this disorder are listed. Our patient has added two abnormalities to this list, pectus excavatum and short first metatarsal bones.

Closed reduction of the knee was attempted at the local hospital shortly after birth, but failed. At our hospital operative treatment was planned for when the respiratory problems were overcome. As a preoperative procedure skin traction was applied. Reduction had almost succeeded when the patient suddenly died. Oki et al. (1976) also reported success in closed reduction of knee dislocation using skeletal traction. Most authors, however, advocate operative treatment and describe surgical procedures (Larsen et al. 1950, Curtis & Fisher 1970).

Table 1. Abnormalities described in patients with Larsen's syndrome (Taken from the reviewed literature). The most typical deformities are in italics.

(A) Abnormalities affecting the musculoskeletal system	
General	*Muscular hypotonia.*
Caput	*Prominent forehead with depressed nasal bridge and widespaced eyes.* Maldeveloped maxilla. Supernumerary incisors.
Vertebral column and rib cage	Spina bifida. Abnormal segmentation. Fusion between atlas and skull. Flattened hypoplastic vertebrae. Pectus carinatum/excavatum.
Upper extremity	*Dislocated elbow. Long, cylindrical ulnar fingers. Spatulated thumbs.* Subluxation in the humeroscapular joint. Hypoplastic, tapered form of the distal humerus. Synostosis between radius and ulna. Multiple carpal ossification centres. Short metacarpals, sometimes with pseudoepiphyses on the bases of the 2nd and 3rd metacarpals.
Lower extremity	*Dislocated hip joints. Anterior dislocation of the knees* *Extra ossification centre of the calcaneus.* Pes equinovarus/valgus. Short 1st metatarsals. Bifid cuboid bone. Subluxation in Lisfrance's joint.
(B) *Other manifestations.*	
Mental development	Retarded in a few patients.
Central nervous system	Gliosis in the cervical medulla (described in a patient with abnormality in the cervical column).
Respiratory tract, oral cavity	*Maldeveloped laryngeal cartilage and tracheal rings.* Cleft palate/uvula.
Genito-urinary tract	Undescended testicles

Micheli et al. (1975) reported three cases of Larsen's syndrome with bone abnormalities of the cervical spine. In one of these cases spinal instability with damage to the spinal cord was considered to be the factor causing a fatal outcome. In our case no spinal abnormalities were found. The attacks of respiratory failure most probably were caused by the maldevelopment of the larynx and trachea. In the medical care of patients with Larsen's syndrome the respiratory problems must be given special attention.

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