Acta orthop. scand. 49, 582–588, 1978

LARSEN'S SYNDROME

Report of Three Cases in the One Family, Mother and Two Offspring

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Three cases are presented of the very rare Larsen's syndrome, being only the second report in the literature of concomitant involvement of parent (the mother in this instance) and offspring (two children) (see also Haberman et al. 1976).

The only therapeutic procedure that is applicable, i.e., surgery, has given only mediocre results over an evolutive period of 8 to 9 years (in case C).

We have not been able to demonstrate analytical or histological abnormalities that might provide a diagnostic clue, so that the aetiopathogeny of the disorder remains unknown.

Key words: Larsen's syndrome; articular luxations; heredity

Accepted 3.iv.78

Since 1950, when Larsen and co-workers described as a nosological entity the group of signs and symptoms constituting this disorder, very few cases have been reported in the literature, and very few variations have been added to the original clinical, analytical and radiological description.

As regards the aetiopathogeny of the disorder, the majority of authors (Almquist et al. 1969, Boni et al. 1974, Steel & Kohl 1972) concur that it must be a generalized mesenchymal alteration. This opinion, however, has not been proven to date.

The possible hereditary character of the disorder has also been the subject of discussion, and the cases reported have been inconclusive in this respect. Some do not show a hereditary trend (Larsen et al. 1950, Lee 1973), some exhibit recessive heredity (Curtis & Fisher 1970, Steel & Kohl 1972, Latta et al. 1971), and some show definite dominant heredity (McFarlane 1947).

In the cases we now present there is no doubt about the hereditary characteristics, but it has not been possible to determine the responsible gene and the type of heredity.

CASE REPORTS

Case A: M.I.A.A., female, 34 years old, born normally at term after an uneventful pregnancy. Defects observed since her birth. No familial history. No familial consanguinity. Husband completely normal. Two living offspring (Cases B and C); one unaffected child died at birth.

Examination

Typical facial features (Figures 1 and 2).

Shoulder: good mobility, no pain.

Elbows: Complete flexion. Bilateral limitation of extension of 20 to 30°. Bilateral luxation (Figure 3).

Hands: Short fingernails, otherwise normal.

Vertebral column: Moderate right dorso-lumbar scoliosis (sleeps with a stiff board under the mattress).

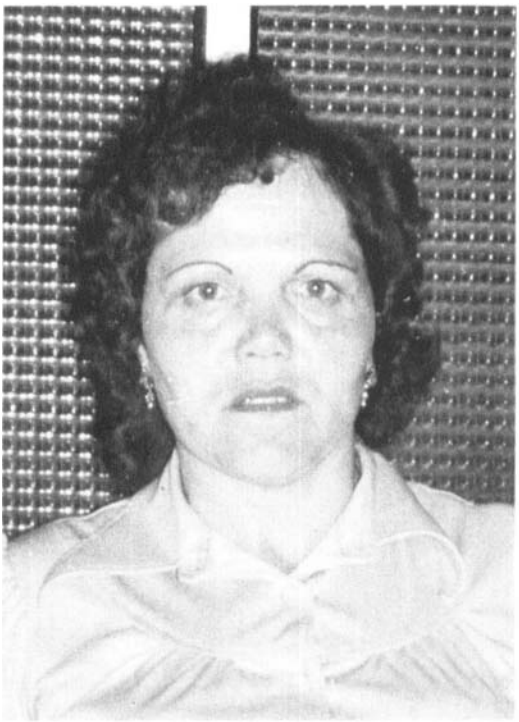

Figure 1. Case A. Typical facial features (nasal depression, hypertelorism, frontal prominence, ample cranium).

Figure 2. Case A. Lateral view of facial features.

Hips: Bilateral luxation (Figure 4), no pain. Acceptable mobility, flexion to 90° on the right and to 80° on the left. External rotation deformity in the left hip; unlimited marching radius.

Knees: On the right side, good mobility without deformities. On the left side, complete extension, flexion to 10°, rotation of tibia on femur with luxation (Figure 5). Bilateral patellar hypoplasia.

Feet: Left pes equino-varo-adductus; right pes plano-valgus. Both feet painless.

Vertebral column: Normal save for discrete dorso-lumbar scoliosis.

Hips: Good mobility, flexion to 100° bilaterally. No contractures or deformities. At present, bilateral subluxation (Figures 8 and 9).

Knees: No deformities and good mobility on the right side. On the left side, flexion to 25° and complete extension.

Feet: Right pes plano-valgus; left pes equino-varo-adductus with marked deformity (Figure 10).

Case B: J.A.P.A., male, 14 years old; born at term, forceps. Defects observed since birth. One parent affected (Case A).

Examination

Typical facial features (Figure 6).

Shoulders: Limitation of mobility, no luxation.

Elbows: Bilateral contracture in 90° flexion. Flexion of 60° in the right elbow and 25° in the left elbow. Bilateral luxation since birth (Figure 7).

Hands and wrists: No deformities, good mobility.

Neck: Slight left torticollis.

Case C: M.I.P.A., female, 11 years old; born at term, breech presentation. Deformities observed since birth. Sister of case B and daughter of case A.

Examination

Typical facial features (Figure 11).

Shoulders: Normal.

Elbows: Bilateral limitation of extension by about 5 to 10°. Bilateral luxation of caput radii.

Hands: Normal.

Neck: Right moderate torticollis.

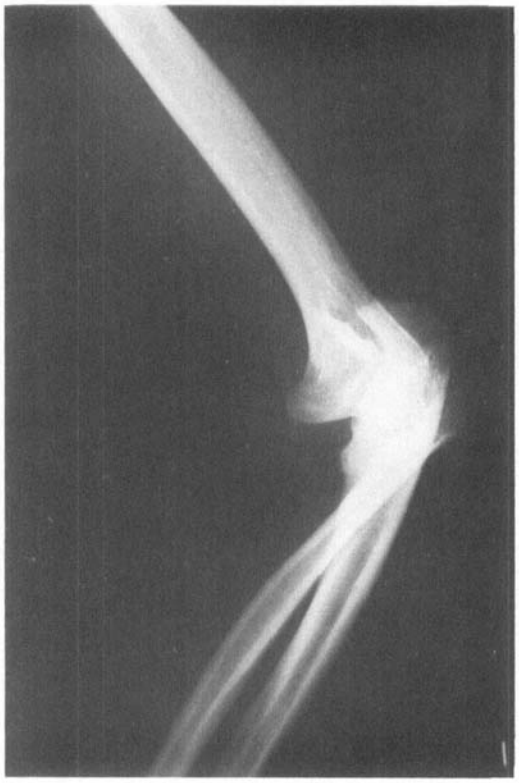

Figure 3. Case A. Left elbow. Superior cubito-humeral and radio-cubital luxation. The disorder is bilateral.

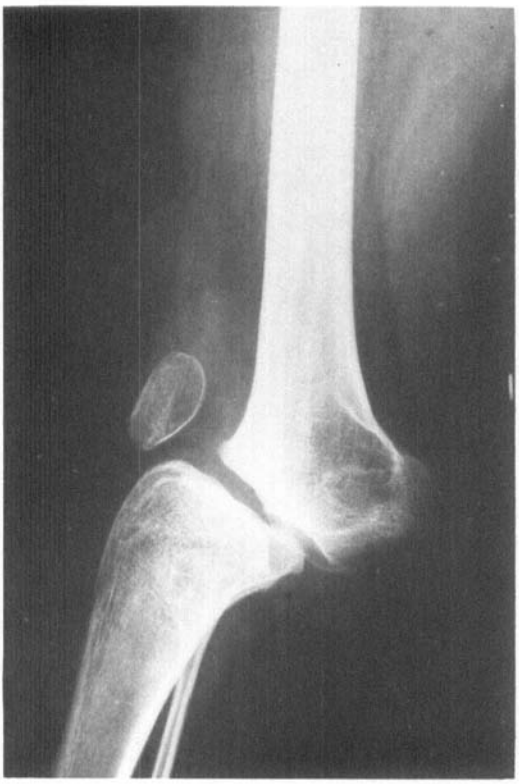

Figure 5. Case A. Left knee. Femoro-tibial luxation.

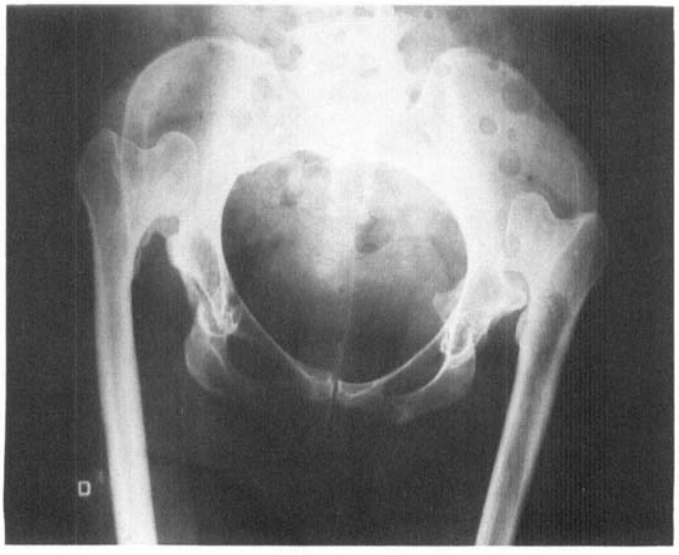

Figure 4. Case A. Hips. Bilateral luxation.

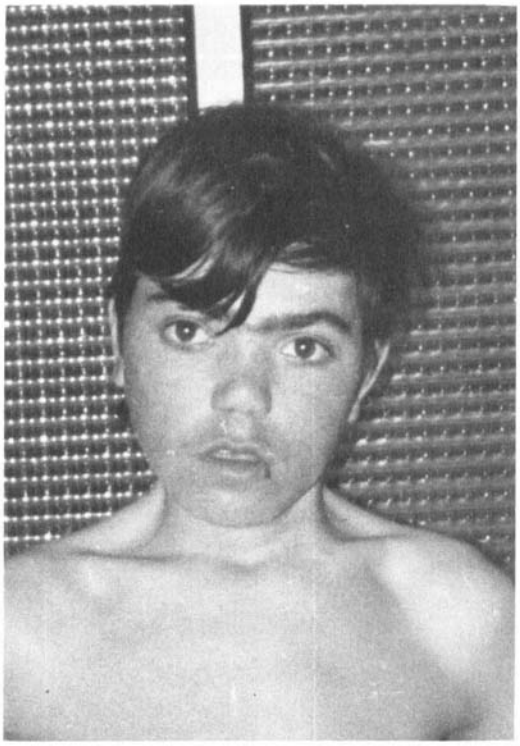

Figure 6. Case B. Typical facial features. Discrete left torticollis.

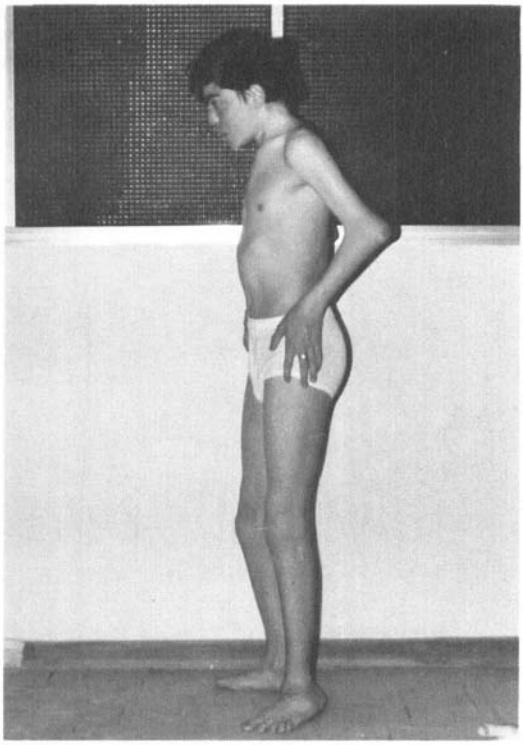

Figure 7. Case B. Bilateral flexion contracture of the elbows and visible deformities of the left foot.

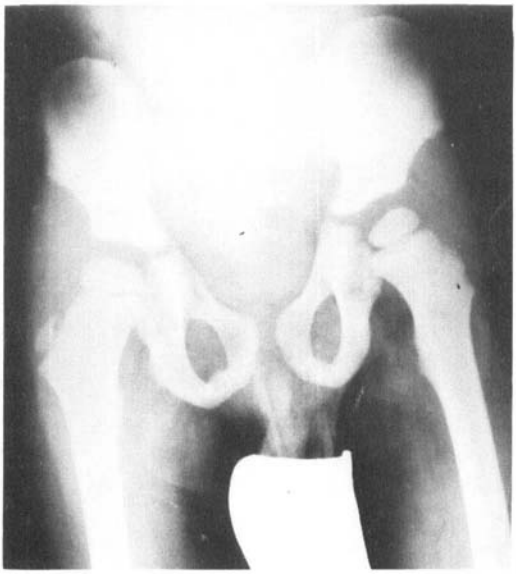

Figure 8. Case B. Hips (1966, age 4 years).

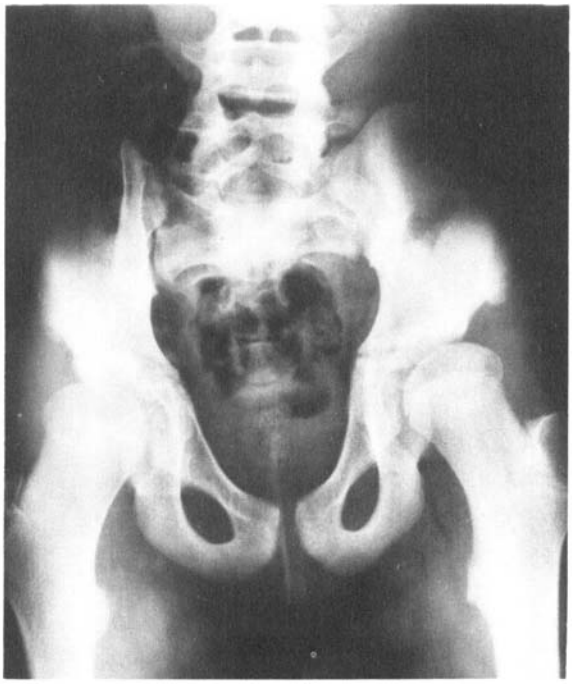

Figure 9. Case B. Hips at present. Bilateral subluxation.

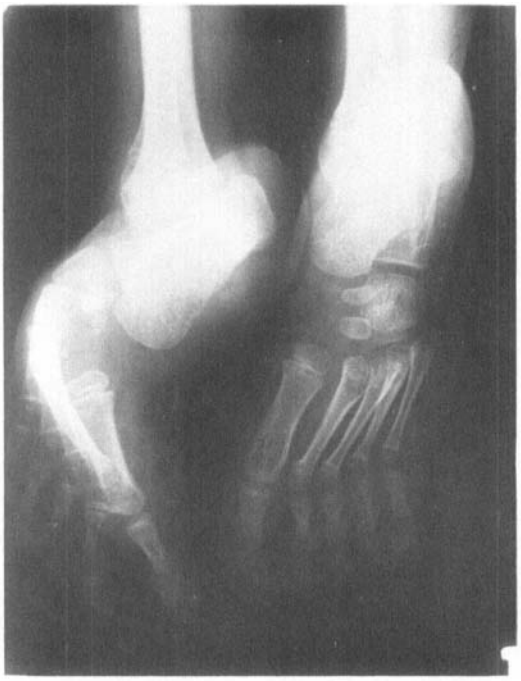

Figure 10. Case B. Feet at present. Marked deformities.

Figure 11. Case C. Typical facial features. Marked right torticollis.

Vertebral column: Normal save for slight right dorsal structural scoliosis.

Hips: Bilateral luxation. Flexion contracture of 20° bilaterally. Limited mobility with flexion to 70° on the right and 80° on the left.

Knees: On the right side, flexion contracture of 20°, flexion to 20°. On the left side, complete extension, flexion to 15°. Discrete deformities.

Lower extremities: Dismetry with shortening by 2.5 cm of the left lower extremity.

Feet: Right pes plano-valgus, left pes equino-varo-adductus.

SURGICAL PROCEDURES

Case A

On the left foot. The patient does not remember date or type of procedure.

Case B

At 18 months, on the left foot; at 24 months, on the right foot; and at 36 months, on the left knee.

All these procedures were carried out prior to the patient's first visit to our hospital, so that we lack all data. In any case, they must be considered as failures, on the evidence of the present state of the areas involved.

At age 6 years: Sternocleidomastoideal tenotomy because of congenital left-sided torticollis. The torticollis persists at present, although it is markedly reduced.

Case C

At age 12 months: Right knee, open reduction of the luxation, stabilization with Kirschner wires, and lengthening of the quadriceps femoris; thereafter, postural plaster cast. On the left knee, a similar procedure save for lengthening of the quadriceps, which was not carried out. At present, there is no luxation (which was so evident before surgery), although discrete postural defects persist and there is very limited mobility.

Table 1. Typical deformities

	Case A	Case B	Case C
Face and cranium			
Hydrocephalus	–	–	–
Palatal defect	–	–	–
Nasal depression	+	+	+
Hypertelorism	+	+	+
Frontal prominence	+	–	–
Articular luxations			
Knees	+	+	+
Hips	+	Subluxation	+
Feet	+	+	+
Shoulders	–	–	–
Elbows	+	+	Subluxation of the caput radii
Wrists and hands			
Multiple carpal ossifying nuclei	+	+	+
Interphalangic luxations	–	+	–
Other anomalies (supernumerary digits, syndactyly, "delta" phalanges)	–	–	–
Vertebral column			
Scoliosis	+	±	+
Segmental anomalies (spina bifida)	+	+	+
Neurologic disorders	–	–	–
Others			
Double ossification nucleus in the calcaneum	Not able to be evaluated due to definitive fusion	+	+
Hyperlaxitude	–	–	–
Mental retardation	–	–	–

+ yes, or present
– no
± discrete

At age 2 years: Right hip, open reduction followed by pelvi-pedic plaster cast. Reduction was impossible, and the luxation persists to date, with deformity of the femoral caput, but with acceptable function.

At all surgical procedures, the pathological study of the soft tissue samples was inconclusive as to diagnosis.

LABORATORY RESULTS

Case A

Blood (white and red cell count, differential white cell count, erythrocyte sedimentation rate) normal. Urine analysis and urinary sediment without pathological findings.

Urinary mucopolysaccharides 26 U c.p.c. per gramme of creatinine (normal). Blood biochemistry in the SMA 12 analytical series (transaminases, lactic dehydrogenase, alkaline phosphatase, albumin, bilirubin, total proteins, cholesterol, calcium, inorganic phosphorus, uric acid, blood urea nitrogen, glucose) within normal ranges. Karyotype 46 XX (normal).

Case B

Blood, urine and urinary sediment (as above) normal; blood biochemistry (as above). Alkaline phosphatase 204 mU per millilitre, inorganic phosphorus 5.6 mg per 100 millilitres; the remainder of values within normal ranges. Urinary mucopolysaccharides 147.2 U c.p.c. per gramme of creatinine. Karyotype 46 XYq (normal).

Case C

Blood, urine and urinary sediment (as above) normal; blood biochemistry (as above). Alkaline phosphatase 147 mU per millilitre, inorganic phosphorus 5.5 mg per 100 millilitres; the remainder of values within normal ranges. Urinary mucopolysaccharides 76.3 U c.p.c. per gramme of creatinine. Karyotype 46 XX (normal).

DISCUSSION

In the cases presented here there is no doubt as to the presence of hereditary factors which are not always present in this syndrome. However, the analytical studies performed yielded normal results, as in practically all cases in the literature, so that the aetiopathogeny of the disorder remains unknown.

It is interesting to point out the absence of pain or neurological signs in spite of the very marked morphological alterations.

Furthermore, ambulation is no problem in the three cases presented; crutches, canes or other aids are not required. The marching radius is not limited.

Surgery has only a relatively limited role in the treatment of this disorder. Apart from the technical difficulties involved, recurrences and residual joint rigidity are frequently encountered. We are therefore faced with a nosological entity presenting great difficulties from the viewpoint of medico-surgical treatment.

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