Cloverleaf skull associated with unusual skeletal anomalies

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Abstract. A male infant with cloverleaf skull and multiple other birth defects born to unrelated, healthy, young parents is presented. Radiologic findings in addition to the cloverleaf skull configuration included short, wide clavicles, winged scapulae, unusual shapes of ribs with abnormal spacing between them and with prominent costovertebral junctions, and widely separated ischia. Ulnae appeared angular with probable fusion to the midportion of the radial bones bilaterally. There was polydactyly of the hands and feet with grossly abnormal metacarpal and metatarsal bones. Skeletal maturation was normal. Computed tomography of the skull showed dilated lateral and third ventricles as well as agenesis of the corpus callosum. The mother denies any teratogenic exposure during the pregnancy. The findings in this infant do not seem to fit into any previously described syndrome.

The cloverleaf skull (CS) syndrome is an etiologically heterogenous entity occurring as an isolated anomaly as well as in association with various pathologic states such as thanatophoric dysplasia and Pfeiffer syndrome [1]. Other conditions with CS include amniotic band, Antley-Bixler, Apert, Crouzon and Carpenter syndromes [1]. Its occurrence in a patient with trisomy 9 has also been reported [2]. We would like to report a patient with cloverleaf skull syndrome who had other skeletal findings that do not fit, to the best of our knowledge, into any previously described syndrome.

Case report

The patient was first seen at age 3 months. He was born to a 29-year-old mother after a full-term pregnancy which was complicated with an urinary infection treated with antibiotics. Delivery was by C-section due to breech presentation. The father was 26 years of age. Consanguinity between the parents was denied. Both parents were in good health except for obesity. The family history showed that on the mother's side there was a first cousin with Down's syndrome and another cousin has epilepsy. The mother's aunt is known to have hypoglycemia while her brother has chronic seizures of unknown etiology. On the father's side, a cousin has severe mental retardation for unknown reasons. The mother has an 8-year-old daughter which was the product of a previous marriage. Finally, she had three spontaneous abortions, all of them taking place at about the fourth week of pregnancy.

Fig. 1. General appearance of the child at 1 year of age
There was no teratogenic exposure during these pregnancies. The birth weight was 3.3 kg and immediate postnatal period was uneventful.

Physical examination showed an unusual looking infant with cloverleaf skull deformity (Fig. 1). The forehead was prominent and the upper part of the skull was tower-like. His face showed marked asymmetry, the left side being larger than the right. The mouth was almost circular in shape and was kept open continuously. He had sunken eyes and the eyelids showed anti-mongoloid slant and blepharophimosis which was more severe on the left. He had a beak-like nose with a depressed nasal bridge. The cheeks were quite prominent with extremely low-set, malformed, and small ears. His chest was narrow and showed a protrusion involving the 8th or 9th rib close to the sternum on the left. Malformations of the extremities were mirror images of each other. Both arms were kept in flexion over the elbows with marked webbing. The forearms appeared to be rather short. He had post-axial polydactyly bilaterally with seven fingers on each hand. The thumb and index fingers were fused with a single fingernail. The index finger was short bilaterally and consisted of only two phalanges. The third and fourth fingers were kept in flexion. The sixth and seventh fingers also showed syndactyly. The abdomen was essen-