Roentgenologic findings of the hydrolethalus syndrome

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Abstract. The hydrolethalus syndrome is an autosomal recessive malformation syndrome which has been recently described in Finland. The name hydrolethalus refers to its main findings, namely polyhydramnios, hydrocephalus and lethality. The patients are either stillborn or die soon after birth. The typical roentgenologic findings are hypoplasia of the tibia associated with the anomalies of the respective bone ray, e.g. metatarsus primus varus atavisticus, hallux varus or hallux duplex varus and hydrocephalus with extreme micrognathia and a specific midline defect of the occipital bone.

Key words: Hydrocephalus – Occipital bone defect – Micrognathia – Polydactyly – Lethal – Hydrolethalus – Skeletal malformation

Roentgenologic findings

The skeletal roentgenologic findings are summarized in Table 1. Hydrocephalus was a consistent finding, and although the degree varied it was severe in most cases. In four cases the head was punctured during delivery and the skull bones were overlapping associated with a midline defect of the occipital bone posterior to the foramen magnum, forming together with the latter a “keyhole” shaped defect in the base of the skull. All had severe micrognathia. Abnormal larynx or trachea, defective lobation of the lungs and abnormal genitalia were seen in six cases. Four had small deepset eyes.

Preaxial polydactyly in the feet was seen in four cases, postaxial in the hands in three cases. Cases 5 and 10 were from the same family. The latter was an induced abortion after ultrasound diagnosis of the hydrocephalic fetus.

Material

The anteroposterior and lateral whole body postmortem roentgenograms of eleven Finnish cases of hydrolethalus syndrome provide the material of the present study. The clinical features of cases 1–4 have been described earlier and they are cases 4, 8c, 13 and 18b, respectively, in the paper of Salonen et al. [1]. All seven new cases had the characteristic phenotype of the syndrome (Fig. 1) and typical findings at the autopsy. All had hydrocephalus...
after delivery. Hydrocephalus was consistently associated with a midline defect of the occipital bone. The bulging margins of the defect had a specific appearance in the lateral roentgenogram of the skull (Fig. 2b). Another consistent skull finding was severe micrognathia.

The upper extremities were short in seven cases (Fig. 3). Four cases had bilateral postaxial hexadactyly of the hands. The lower limbs showed abnormalities in all cases except one (Case 8). The proximal hypoplasia of the tibia could be seen in seven cases (Fig. 3). Case 10 had aplasia of the tibiae and two cases had normal tibiae. A short tibia was frequently associated with anomalies of the respective bone ray, e.g., short first metatarsal bone, double hallux varus and short hallux varus (Fig. 4). Case 3 had unilateral double hallux with normal sized tibia and case 7 had short tibiae without hallux anomalies. In some cases cutaneous syndactyly could be seen between the doubled halluces.