THE SKULL IN ACHONDROPLASIA

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The growth disorder in achondroplasia results from abnormalities of endochondral bone formation. Cranial abnormalities originate from the occipital bone, the only region where enchondral bone is formed.

The occipital bone consists of a) the basioccipital, b) the lateral parts, c) the planum nuchae, and d) the planum occipitale. The latter derives from membranous bone, the remaining three from endochondral bone formation.

Comparison of an achondroplastic with a normal skull shows 1) that the endochondral-derived bones (a-c) are stunted, shorter and narrower and 2) that the squama of the occipital bone is horizontalized (8). As a consequence the skull base is longer and narrower, the posterior fossa is distorted (13) and the occipital foramen is smaller than normal (5).

Narrowing of the occipital foramen has been associated with compression of the upper cervical spinal cord and lower brainstem with ensuing respiratory complications, hypoxemia and death (3,4,9,12,14, 15,20).

The abnormalities at the skull base have also been claimed to obstruct cerebrospinal fluid pathways (1). Accordingly, cisternography showed delayed CSF circulation (16).

This finding was not confirmed by others (13) who suggest that CSF flow is normal but that venous flow is impaired. They claim that a stenosis of the sigmoid sinuses leads to a retrograde increased venous pressure thought to be caused by an abnormally small and distorted jugular foramen. Unfortunately, the jugular foramen has never been adequately investigated in achondroplasia. The only patient in whom narrowing of these orifices was anatomically demonstrated had thanatophoric dysplasia with a cloverleaf skull but not achondroplasia (18).

Though we do not know the exact pathomechanism, there is no doubt that the bony abnormalities and ensuing impairments lead to the intracranial accumulation of CFS with dilatation of the ventricles and cortical sulci (Fig. 1).
The accumulation of fluid leads to an increase of the intracranial pressure. This can be concluded from the increasing head size which is present in the large majority of patients with achondroplasia (6). It is confirmed by direct measurements (Fig. 2).

There is little doubt that long-lasting elevation of the intracranial pressure has adverse effects on the brain, once compensatory mechanisms are exhausted. In the infant it causes destruction of parenchyma with neurologic and mental impairment (7).

In achondroplasia, the hydrocephalus stabilizes spontaneously in most cases by the age of 2 to 3 years. This is concluded indirectly from the flattening of the head growth curves and the absence of clinical signs of increased intracranial pressure.

The question remains if and how much cerebral damage occurs during the period of increased intracranial pressure. In the absence of pathohistologic studies the question can be answered only by studies comparing brain function of children with and without achondroplasia, i.e. by the evaluation of their neurologic and mental function.

Studies of the early motor development of achondroplastic children show a marked retardation of gross motor abilities (17). This is in