Vascular Malformations of the Brain in Achondroplasia

Case Report

By

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With 3 Figures

Summary

A further case of intracranial haemorrhage in achondroplasia is reported. At necropsy, an intracerebral haematoma communicating with the left temporal horn was found. Histological examinations revealed the presence of a cavernous angioma. The literature is reviewed.

Keywords: Vascular malformations; achondroplasia.

Achondroplasia, a chondrodystrophy inherited as an autosomal dominant gene, is frequently associated with well-known neurological disorders, mainly related to hydrocephalus, megalencephaly, brain heterotopias, and compression and malformations of the spinal cord and the cauda equina. To the best of our knowledge, only two cases of association with vascular anomalies of the brain have been reported. In the present paper, a further case is presented.

Case Report

A 56-year-old achondroplasic dwarf presented with sudden headache and vomiting followed by gradual loss of consciousness. On physical examination, the unconscious patient showed marked nuchal rigidity. The pupils were equal in size and reacted to light. The corneal reflexes were present on both sides. Right hemiplegia was apparent, including homolateral facial weakness. The deep tendon reflexes were absent on the right. A spinal tap revealed bloody cerebrospinal fluid. Plain X-rays of the skull were normal. Serial left carotid angiograms demonstrated moderate displacement of both the anterior cerebral and the lenticulostriate arteries, without any evidence of vascular malformations (Fig. 1). In spite of intensive care, the patient died three hours later. At necropsy, the brain weighed 1,350 grams.
intracerebral haematoma (Fig. 2) with extension to the left temporal horn was found. Histological examination of specimens taken from the medial wall of the cavity demonstrated the presence of a cavernous angioma (Fig. 3).

Fig. 1. Left carotid angiogram. Anteroposterior view. Subtracted film. The vascular malformation is not recognizable.

Comment

Despite the high incidence of neuropathological lesions in achondroplasia, a condition requiring a neurological evaluation in half of the affected patients 3, 10, association with vascular malformations of the brain results only from two observations 9. In the first case, postmortem examination revealed "an excessive number of abnormal blood vessels within the choroid plexus". In the second, a ruptured saccular aneurysm of the left middle cerebral artery was found. In