Cranial Morphology in the 18p⁻ Syndrome

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Abstract. Three of four recently described children with the 18p⁻ syndrome were reinvestigated using cranial computerized tomography (CCT). More severe deformities were found in the cases with severe cerebral malformation, but there was no correlation with the degree of mental retardation.

Key words: Cranial computerized tomography – Congenital anomalies – Mental retardation – Structural aberrations – Deletion syndromes.

Introduction

Autosomal aberrations in man give rise to malformation syndromes—including craniofacial dysmorphia—and to mental retardation (Schinzel, 1976). In 1976 we described four patients with the 18p deletion syndrome who showed various degrees of head deformity (Faust et al., 1976). We reinvestigated three of these patients using cranial computerized tomography to elucidate the relationship between the external abnormalities and the cerebral morphology. Furthermore we were interested to ascertain whether the degree of mental retardation correlated with the cerebral malformation.

Methods and Material

Patients

Clinical data have been given elsewhere (Faust et al., 1976). Case 1 (D.) exhibited the median harelip and cleft palate typical of holoprosencephaly. Autopsy showed lobar holoprosencephaly.

The three surviving patients (cases 2, 3, 4)—each six years of age—revealed the classical clinical picture of the 18p⁻ syndrome: i.e., microcephaly, midface retraction, hypertelorism, a broad-based flat nose, low-set and protruding ears, a “carp mouth” with down slanting angles, a
low hairline, failure to thrive, and mental retardation. In addition, the head of the third patient (R.B.) was turricephalic with a flat occiput and a protruding forehead. In all three patients, there was retardation in mental development by 20—24 months.

**CCT**

The investigations were performed with a Siretom II (Siemens), using the 129 x 129 matrix. To avoid artifacts caused by movements of the head, the children were sedated with chloral hydrate. No general anaesthesia was necessary.

**Results**

*Patient No. 1 (D.)*

CCT was not performed. The postmortem pneumencephalogram revealed communicating lateral ventricles, absence of the septum pellucidum, and an intra-