UNILATERAL SOMATIC AND INTRACRANIAL HYPOPLASIA

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Introduction

Unilateral somatic and intracranial hypoplasia (USICH) does not represent a true neurocutaneous disorder, but most of its features including unilateral hypoplasia involving the upper and lower extremities, the breast and the trunk, bilateral acral abnormalities affecting especially the middle phalanges, mental retardation and partial epilepsy, overlap with the clinical and imaging findings of other neurocutaneous diseases and/or complex malformation syndromes described in this book, such as oculocerebrocutaneous/ Delleman syndrome (OCCS), encephalocraniocutaneous lipomatosis (ECCL), Proteus syndrome, Parry-Romberg syndrome, scleroderma " en coup de sabre" and Goldenhar syndrome.

Historical perspective

This syndrome was first characterized by Pascual-Castroviejo et al. (1997).

Incidence and prevalence

Even though no new cases have been reported since its first clinical description (Pascual-Castroviejo et al. 1997), we have seen additional patients, all women, with the same features.

Clinical manifestations

The anomalies involve structures of ectodermal and mesodermal origin. Main clinical features are: unilateral hypoplasia that involve the upper and lower extremities, breast and trunk (Fig. 1), bilateral acral abnormalities affecting especially the middle phalanges (Fig. 2), mental retardation and partial epilepsy. Patients may present with hemifacial hypoplasia of the contralateral side due to the early and severe lesion of the affected cerebral hemisphere. Specific defects include several malformations ipsilateral to the hypoplastic hemibody such as unilateral cerebral brainstem (Fig. 3) and/or cerebellar hypoplasia, enlargement of the corresponding lateral ventricle with cortical polymicrogyria (Fig. 4), agenesis of the corpus callosum, microphthalmia and cataracts. All intracranial vessels on the affected side appear hypoplastic as well (Fig. 5). Despite phenotypic overlaps between USICH and some other neurocutaneous diseases described in this book (see above), the specific developmental malformation consisting of hypoplasia of the entire hemibody in women suggests that this represents a new entity.

Molecular genetics and pathogenesis

USICH malformations correspond to different embryological periods. Hypoplasia of cerebellar hemisphere has been described in association with facial hemangioma or vascular malformation, mostly on the ipsilateral side (Pascual-Castroviejo 1978; Pascual-Castroviejo et al. 1985, 1986), and this occurs since $4^1/_2$ weeks of gestational age. However, the cerebellum has the longest period of embryological development as compared to any other major structure of the brain. Congenital and acquired vascular etiologies have been suggested (De Souza et al. 1994, Granados-Alzamora et al. 2003, Pascual-Castroviejo

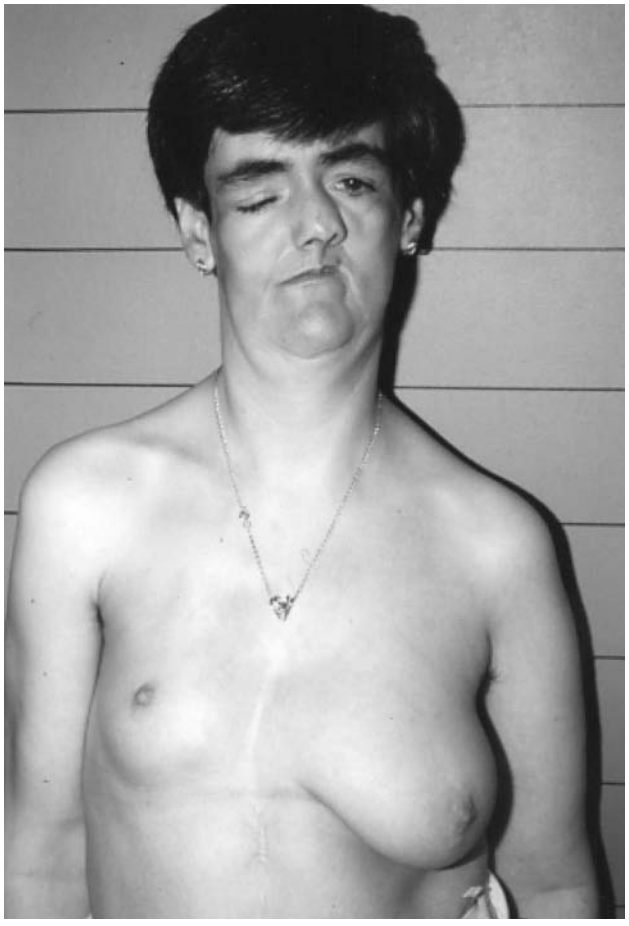

Fig. 1. Photograph of a patient at 30 years of age shows palpebral ptosis and breast hypoplasia on the right side and left facial atrophy.

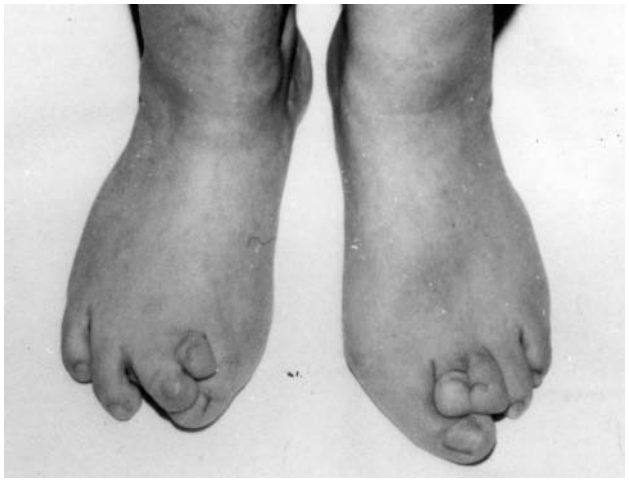

Fig. 2. Right foot hypoplasia and bilateral toe deformities.

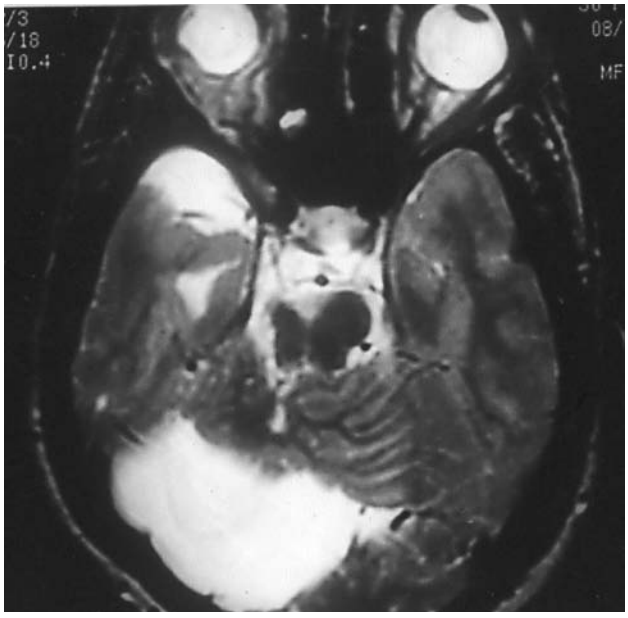

Fig. 3. Axial T_2-weighted axial MR image (400/88/2) of the upper part of the posterior fossa, the lower part of the middle fossa and the orbits shows hypoplasia of the right cerebellar hemisphere, of the right part of the mesencephalon, of the right temporal lobe and of the right eye.

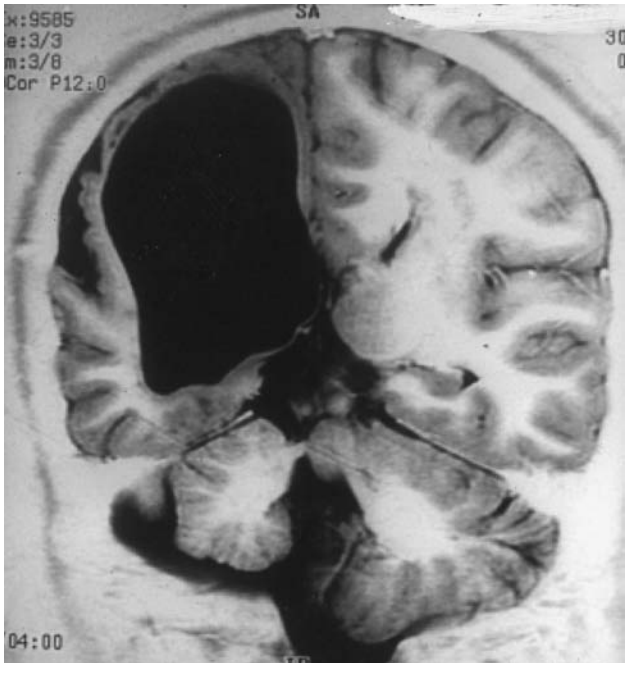

Fig. 4. Coronal MRI (600/48/3/300) of the middle-posterior region of the brain discloses dilatation of the right cerebral ventricle with very thin cerebral parenchymal cortical polymicrogyria, and severe hypoplasia of the right cerebellar hemisphere.

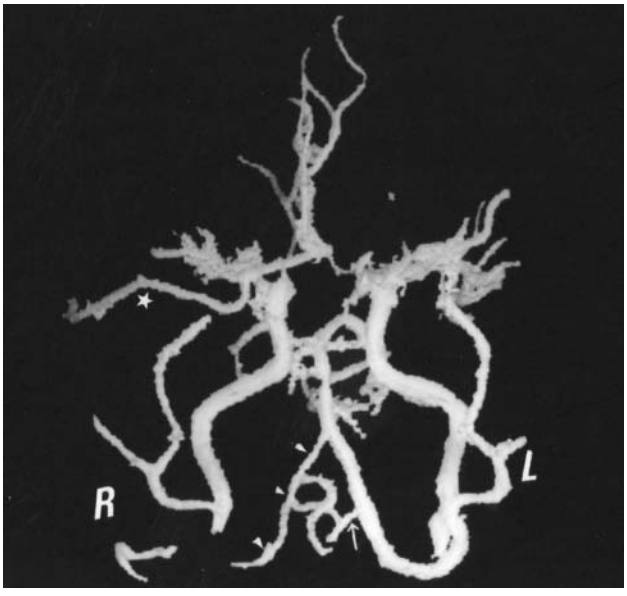

Fig. 5. Three-dimensional arterial phase contrast RM angiogram (48/6.9/1) in coronal view reveals the presence of only one posterior inferior cerebellar artery (PICA) which originates from the left vertebral artery of normal size (arrow), while the right vertebral artery is hypoplastic (arrowheads), and a low hypoplastic right middle cerebral artery (star).

et al. 1975, Pascual-Castroviejo 1978a). The period of cortical plate formation begins with the first wave of migrating neuroblasts from the subventricular region, at about 7–8 weeks of gestation (Sarnat 1992). The corpus callosum, however, starts to develop at 10–11 weeks and agenesis may cause changes in gyration (Muller 1990). Appearance of the middle phalanges in the hands and feet is at 12–14 weeks of gestation (Wood and Dimmick 1992). Ocular and palpebral paralysis with cataract homolateral to the remaining affected structures may be associated (likely secondary) to the cerebral hemisphere and brainstem lesions.

Treatment

The treatment of this syndrome is symptomatic.

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