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REVIEW

Malformations of cortical development of the human brain: A pictorial essay

Malformations du développement cortical du cerveau humain : une revue iconographique

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Summary During development in utero of the human brain, an error in one or more of the orderly processes of neuroblast proliferation and differentiation, neuroblast migration and cortical organization may result in disordered neocortical development. Nowadays, the consequent malformations of the cerebral cortex and associated structures are detectable on pre- and postnatal examination with growing frequency, thanks to the evolution of modern imaging modalities. In particular, magnetic resonance imaging (MRI), due to its excellent contrast differentiation and multiplanar capabilities as well as the development of even newer techniques, such as diffusion tensor imaging and spectroscopy, has surpassed all other forms of imaging for the thorough exploration and analysis of congenital anomalies of the central nervous system. These malformations comprise a heterogeneous group of conditions in terms of both the timing and etiology of the developmental aberration as well as the resulting morphological phenotype, including epilepsy, developmental delay/intellectual disability and focal neurological deficits. This study briefly presents some typical examples of congenital malformations of cortical development of the human brain that are encountered in practice. It is our belief that familiarity with the MRI presentations of these conditions can be of considerable value for adequate disease management and genetic counseling.

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Introduction

In ancient Sparta, there was a cruel tradition of killing infants who were deemed deformed or mentally retarded by throwing them from Mount Taygetos into a deep chasm called Ceadas. Luckily, modern societies have put such practices behind them. Nowadays, science provides the means by which to diagnose and treat many conditions that just a few decades ago would have branded such sufferers as ''outcasts''.

Considerable progress has been made especially in the study of malformations of cortical development (MCD) of the human brain, which can result in developmental retardation, epilepsy and focal neurological signs. Its causes include an error in one of the following principal procedures: cell proliferation and apoptosis; neuronal migration; and cortical organization. In addition to the classical magnetic resonance imaging (MRI) sequences, which offer only morphological analyses, the development of new sequences, such as diffusion imaging and spectroscopy, enable monitoring of the functional, biochemical and metabolic development of the human brain.

A knowledge of and familiarity with the MRI manifestations of MCD can be particularly useful for the early detection, evaluation, and improved patient treatment and disease management of especially those with MCD, which can be potentially ''curable'', as well as for genetic counseling of affected families.

Imaging techniques

The present report presents some of the most common MCD encountered during routine MRI at our institute, performed with 1.5 and 3 T Siemens and Philips MRI devices (Siemens, Erlangen, Germany; Philips Achieva, Eindhoven, The Netherlands). MRI is currently the modality of choice for visualizing the suspected congenital anomalies of the brain on pre- and postnatal examination. In most of our cases, the patients were children who presented with seizures and—depending on the degree of malformation—psychomotor retardation. The initial study was usually performed while the symptoms were present. Today, high field-strength units of 3 T with 32-channel sensitivity-encoding head coils are available in everyday practice for the optimal study of MCD, and provide high-resolution images within a reasonable acquisition time [1].

The present study implemented an adequate acquisition time to achieve motion-free images and higher signal-to-noise-ratios, as well as millimetre-section thicknesses, to provide high resolution and the possibility of multiplanar reconstruction. High-quality multiplanar images are essential for adequate analysis of lesions and more accurate determination of their exact borders as part of the preoperative workup in those subjects suitable for surgical treatment.

The technical protocol used included fat-saturated T2-weighted (FST2), three-dimensional T1-weighted (3DT1), and new techniques such as diffusion tensor imaging (DTI), fiber tractography (FT), spectroscopy and arterial spin labeling blood flow (ASL) MRI sequences to localize the precise defective fibers and zones of hypo- or hyperperfusion, depending on the ictal and postictal time of acquisition, respectively. These latest techniques can be obtained without contrast administration, clearly an advantage when imaging children.

In addition, different reconstruction algorithms have been used by experienced neuroradiologists to better visualize the MRI manifestations of the different MCD. Three-dimensional (3D) reconstructions with multiplanar reconstruction (MPR) and maximum intensity projection (MIP) algorithms in selected section thicknesses are easily feasible in approximately 10 min. In cases where the suspected anomaly reaches the surface of the cerebral cortex, curvilinear reconstructions can also be made in no more than 10 min. In addition, DTI reconstructions are possible in approximately 10 min, and are essential for presurgical planning and pinpointing pathological fibers, thereby helping to lower the risk of postoperative deficits. ASL reconstruction is automatically provided for by MRI software.

Description

During week 5 of gestation, a proliferation of neuroblasts—called the ''germinal matrix''—develops in the subependymal layer of the lateral ventricles. During week 7 of embryogenesis, the hemisphere wall consists of the inner germinal-matrix layer and a superficial acellular zone. After approximately eight weeks of gestation, the cortical plate consists of three to five rows of cells that have migrated radially out from the germinal matrix. The six-layer neocortex emerges from this pattern by the sixth or seventh month of gestation [2].

Based on these pertinent aspects of central nervous system (CNS) embryology, the 2005 classification of MCD distinguishes disorders of cell proliferation, migration and cortical organization, classifying them according to the developmental stage during which disruption is considered to be the cause of the anomaly [3].

According to this classification scheme, there are four groups of malformations. Group I comprises disorders with decreased/increased proliferation or the proliferation of abnormal cells, and includes microcephaly, focal cortical dysplasia with balloon cells, hemimegalencephaly, tuberous sclerosis, dysembryoblastic neuroepithelial tumor and ganglioglioma/gangliocytoma. Group II includes malformations due to abnormal neuronal migration, such as lissencephaly, dystroglycanopathy and heterotopia. Group III comprises disorders due to abnormal cortical organization (including late neuronal migration), and also includes polymicrogyria, schizencephaly, cortical dysplasia without balloon cells and microdysgenesis. Finally, group IV includes all other MCD that are not otherwise classified, such as mitochondrial disorders and sublobar dysplasia (Table 1).

The present study follows this classification system, although MCD classification remains a challenge for both neurologists and neuroradiologists as, in many cases, multiple disorders are coexistent and, in others, the etiology appears to be heterogeneous [4,5].