CONGENITAL BRAIN MALFORMATIONS: NEUROIMAGING REVIEW

2017 CNSF Resident Review Course

Tuesday, 20 June 2017

Congenital cerebral malformations encompass a wide spectrum of conditions that can affect the developing brain and result from absence or disruption of normal developmental processes. A general approach is to consider malformations resulting from either abnormal gene expression, or from injury to the brain at a time when a gene is expressed.

Malformations of cortical development represent the largest component of brain malformation syndromes, and are increasingly being discovered on imaging examinations of children and young adults with developmental delay and intractable seizures.

The most commonly used classification system of congenital malformations of cortical development is based upon the developmental steps at which the process is first disturbed. A more detailed description of the many possible syndrome variants requires a level of developmental and genetic understanding that is beyond the purpose of this lecture, and the reader is referred to the excellent works of Barkovitch et al (2012).

From an imaging viewpoint, the three main categories of cortical malformations to consider are: 1) abnormal neuronal and glial proliferation or apoptosis, 2) abnormal neuronal migration, and 3) abnormal late migration or cortical organization. Malformations of abnormal neuronal and glial proliferation or apoptosis include microcephaly, focal cortical dysplasia with balloon cells, and megalencephalies. Malformations due to abnormal neuronal migration include heterotopia, classic and incomplete lissencephaly, subcortical heterotopia, and cobblestone malformations. Malformations secondary to abnormal post-migrational development include polymicrogyria, schizencephaly, and focal cortical dysplasia without balloon cells.

There are many excellent published and on-line educational resources that already discuss and provide detailed imaging descriptions of the various congenital malformations. This lecture will discuss a basic radiologic approach to interpreting imaging exams of pediatric patients with brain malformation syndromes, focusing on the more common and important congenital malformations of cortical development.

SUGGESTED RESOURCES:

Barkovitch, A.J., 2007. Diagnostic Imaging: Pediatric Neuroradiology. 1st ed. Salt Lake City, Utah: Amirsys.

Kuzniecky, R.I. and Jackson, G.D., 2005. Magnetic Resonance in Epilepsy. 1st ed. New York, NY: Elsevier.

Barkovitch, A.J. et al. A Developmental and Genetic Classification for Malformations of Cortical Development: Update 2012. Brain (2012); 135(5): 1348-1369.

U.S. National Library of Medicine National Institutes of Health - Genetics Home Reference (https://ghr.nlm.nih.gov).