Editorial

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Pachygyria with Mental Retardation and Seizures

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Editorial

Pachygyria is neuronal migration disorder with thick convolutions on cerebral cortex which leads to mental retardation and intractable seizures. It is also called as Macrogyria. It is a rare disease and diagnosis is increasing with advances in the imaging techniques. This autosomal recessive neurodevelopmental disorder is characterized by mental retardation, seizures pachygyria and diffuse localization of arachnoid cysts.

It most likely represents a neuronal migration disorder within the lissencephaly spectrum. Lissencephaly, Pachygyria and polymicrogyria are all the results of abnormal cell migration. The abnormal migration is typically associated with a disorganized cellular architecture, failure to form six layers of cortical neurons and functional problems. The abnormal formation of the brain may be associated with developmental delay, seizures and mental dysfunctions. Pachygyria is caused by a breakdown in the fetal neuronal migration process due to genetic or possibly environmental influences. The cerebral cortex will typically have only four developed layers.

One of the best known and most common types of neuronal migration disorders is lissencephaly. Incomplete neuronal migration during the early fetal brain development is the precursor to lissencephaly. Should neurons follow an abnormal migration during development possible cortical malformations include classical lissencephaly and subcortical band heterotopia with an agyria-pachygyria band spectrum.

Most common types of incomplete neuronal migration to the cortex occur during the third and fourth gestational months. The abnormal migration of the neurons causes them to not reach their proper final destinations which results in failure of the sulci and gyri to form. The stage of cortical development at which migration is arrested is directly related to the level of structural malposition. One of the most critical stages in brain development is when the post-mitotic neurons migrate from the ventricular zone to form the cortical plate. Migration arrested towards the later part of development usually restricts the abnormal cell position to the cortex level.

LIS1 is responsible for the autosomal form of lissencephaly. Mutations of the *LIS1* gene are associated with about 80% of those affected with lissencephaly. *LIS1* was the first human neuronal migration gene to be cloned. It is responsible for encoding the alpha subunit of the intracellular Ib isoform of platelet-activating factor acetylhydrolase. It is located on chromosome 17p13.3 and has 11 exons with a coding region of 1233bp. *LIS1* protein appears to interact with tubulin to suppress microtubule dynamics.

The protein is highly conserved and studies have shown that it participates in cytoplasmic dynein-mediated nucleokinesis, somal translocation, cell motility, mitosis, and chromosome segregation. *LIS1* encodes for a 45kDa protein called PAFAH1B1 that contains seven WD40 repeats required for proper neuronal migration. The *LIS1* gene encodes for a protein similar to the β subunit of G proteins responsible for degrading bioactive lipid Platelet-activating Factor (PAF). Because pachygyria which is a structural defect no treatments are currently available other than symptomatic treatments, especially for associated seizures. Another common treatment is a gastrostomy to reduce possible poor nutrition and repeated aspiration pneumonia.

