Case of the Month – January 2014

• 20 year old female presenting with increased seizures. No fever or trauma history.
• Past history of recurrent seizures since childhood and cognitive impairment.
• Investigations reveal:
  – Unremarkable bloodwork
  – No infectious etiology.

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Unenhanced CT of the Head

- What are the imaging findings?
- What is the differential diagnosis? Diagnosis?
Axial and Coronal
Sagittal and Axial
Findings

• Thickened cortex with diminished cortical gyri that demonstrate simplified gyration pattern (pachygyria)
• Shallow sylvian fissures (not shown)
• Lack of white matter arborization and presence of sparse white matter zones
• Periventricular calcifications
• Incidental note of thickened calvarium – perhaps secondary to anti-epileptics
Differential Diagnosis

- Immature brain?
- Lissencephaly?
- Band Heterotopia?
- Other?
Diagnosis

• Findings are most in keeping with **lissencephaly secondary to CMV infection**.

• Although band heterotopia overlaps with lissencephaly there is no evidence of a ‘double-cortex’ sign (abnormally located subcortical grey matter) to suggest this diagnosis.

• Immature brain excluded based on patient’s age (sulci develop at ~40 weeks gestation).
Discussion

• A rare diagnosis which encompasses a wide spectrum of abnormalities

• Characterized by abnormal cortical formation as a result of arrested migration of neurons. This results in a thickened cortex made up of arrested columns of cells and decreased sulci formation. May see microcephaly.

• Periventricular calcifications suggest CMV as etiology

• Patients often suffer from seizures, facial dysmorphism, cognitive impairment, developmental delay.
Discussion

• Pathophysiology:
  – Genetic: mutations which affect the proteins required for migration of neurons resulting in arrest. Genes involved include *LIS1, DCX, RELN, ARX*
  – Acquired: infections (CMV) or toxins (alcohol) result in arrest of neuronal migration

• Treatment: supportive therapy and seizure control
References


• StatDx – Lissencephaly