ACC and Associated Features

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- *MRI features associated with ACC*
- *Clinical diagnoses found in individuals with ACC*
- *Clinical Syndromes in which ACC is a component or a major piece*
- *Your questions!*

The Normal Corpus Callosum

- *Midline structure that connects the two hemispheres*
- *200 million axons cross through this structure*
- *Develops between 10-16 weeks in utero, continues to grow until adulthood*

Planes of View: Head MRI

Adult Female, Normal CC

Complete Callosal Agenesis
Callosal Hypogenesis
Thin corpus callosum

MRI Features that can be Associated with ACC

- *Colpocephaly*
- *Upward displacement of lateral ventricles*
- *Probst bundles*
- *Interhemispheric cysts*
  - ***Type I***
  - ***Type II***
  - ***lipoma***

Colpocephaly

- *Colpo= "hole", cephaly= "head"*
- *This is a posterior enlargement of the lateral ventricles that results from the absence of the corpus callosum*
- *Similar phenomenom is the "steer-horn" shaped lateral ventricles seen more anteriorly.*

Colpocephaly
Upward displacement of anterior lateral ventricles
Probst Bundles

- *Axons that are unable to cross the midline*
- *Turn and run in large bundles longitudinally*
- *Unclear clinical significance, although some hypothesize that presence of bundles correlates with better outcome*

ACC & Probst Bundles
Interhemispheric Cysts

- *These cysts are fluid filled structures, outside of the brain matter proper*
- *Current thinking: cysts develop in lieu of callosum, not that cysts displace callosum*
- *Type I-communicate with ventricles*
- *Type II-not communicate with ventricles*
- *Continuum with lipomas*

Type II Interhemispheric Cyst

Cortex Layering and ACC
Periventricular Nodular Heterotopia
ACC and Gray Matter Heterotopia
Lissencephaly
Lissencephaly with ACC
and Cerebellar Hypoplasia
Simplified Sulcation and
Callosal Hypoplasia
Chiari II Plus
Abnormal White Matter and Subependymal Heterotopia
What other MRI techniques will allow us to understand the nature of ACC?

Can My Child Have ACC and:

- *Autism*
- *Attention Deficit Hyperactivity Disorder*
- *Obsessive Compulsive Disorder*
- *Cerebral Palsy*
- *Mental Retardation*

Autism: DSM IV
Autism: NICHD Definition
ADHD Definitions

- *A learning disability marked by inattention, impulsiveness, a low tolerance for frustration, and a great deal of inappropriate activity*
- *A syndrome (a group of symptoms or signs) that is usually characterized by serious and persistent difficulties resulting in: inattentiveness or "distractibility", impulsivity, and hyperactivity.*

OCD

- *A disorder in which individuals are plagued by persistent, recurring thoughts (obsessions) that reflect exaggerated anxiety or fears*

- *Responds very well to SSRI's supplemented with cognitive therapy*

Cerebral Palsy

- *a broad term that describes a group of neurological (brain) disorders. It is a life-long condition that affects the communication between the brain and the muscles, causing a permanent state of uncoordinated movement and posturing.* CP is the result of an episode that causes a lack of oxygen to the brain.

Why assign these diagnoses?

- *Potentially better understanding of the issues that your child faces*

- *Useful rubric from which to prescribe medications*
- *Helpful for obtaining therapy services*

Why not to assign these Dx's?

- *Allows medical community to lump child together with others, minimizing the effort placed on individual assessment and tailoring of therapy*
- *Stigmatizes the child emotionally, socially*
- *Has untoward insurance repercussions*
- *Leads to a cessation of the diagnostic evaluation*

What does it mean to have a syndromic diagnosis?

- *Depends on:*
  - ***How the diagnosis was made***
    - Molecular genetic testing
    - Precise clinical features
    - Gestault clinical constellation
  - ***What information you want to know that accompanies the diagnosis***
    - Risk of genetic transmission by patient or other family members
    - Range of outcomes for your child
    - Risk of having another affected child
    - Involvement in parent/patient support groups
    - Participate in ongoing research (diagnostic/therapeutic)

Fetal Alcohol syndrome

- *Abnormal development of the fetus and infant caused by maternal alcohol consumption during pregnancy. Features of the syndrome include retarded growth, small head circumference, a flat nasal bridge, a small midface, shortened eyelids, and mental retardation.*
- *World-wide prevalence: 1.9/1,000 births*
- *ACC is a not uncommon brain malformation see in FAS*
- *However the diagnosis can be difficult to make, as features can be subtle*
- *No good evidence that small amounts of EtOH can lead to FAS*

FAS-Facial Features
ACC Syndromes with molecular genetic testing

- *Andermann Syndrome*
- *Mowat-Wilson*
- *ARX related syndromes*
- *L1CAM related syndromes*

ACC syndromes without genetic testing
Conclusions

- *ACC is a radiographic phenotype that has multiple causes*
- *Frequently ACC is associated with other changes in brain development--the clinical significance of some of these associations is known, for others it is being studied*
- *Individuals with ACC often do not fit perfectly into the available clinical diagnoses, there are pros and cons to pursuing these diagnoses*
- *ACC can be part of a "syndrome".*
  - ***Some syndromes are just a constellation of clinical findings that may or may not have a high degree of correlation***

  - ***For cases in which the gene is known, the outcomes observed for the syndrome itself are most important***
- *This talk serves as an introduction to this complex topic--ask lots of questions of your doctors*