HOW TO GET THE MOST FROM YOUR NEUROLOGIST VISIT

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• General issues for your visit with the neurologist
• ACC definitions

Who are Child Neurologists and what do they do?
• MDs who are trained in pediatrics and neurology (should be board certified in child neurology)
• Evaluate and treat children with neurological disorders
• Serve as a gatekeeper for evaluation and therapy (from perspective of insurance company)
• Tests they order and perform: (MRI, EEG, EMG/NCS, Spinal tap, blood tests, etc)
• They work in concert with
  – Neurosurgeons
  – Neuroradiologists
  – Behavioral Pediatricians
  – Psychiatrists and Psychologists
  – Therapists (PT, OT, Speech)

Why would you want to see a Child Neurologist?
• Diagnose condition
• Medical management of symptoms
  – Seizures
  – Behavior
  – Spasticity
• Coordinate care and resources
• Referral to other specialists
• Any unexplained neurological or behavioral problems, particularly if they have newly arisen

Practical considerations for your visit with the Neurologist
• Bring someone with you
  – For moral support, to help you remember what was said, to watch your child during the doctor-parent discussion
• Consider taping the session
  – Or take good notes
• Write your questions down before your visit and accumulate new ones leading up to the next visit
• What else to bring:
  – Results of any prior testing
  – IEP reports (if you would like help here)
  – Family history and photographs

Visit issues: continued
• Ask that all records be given to you as well (consult letters, test results (and actual raw data at times-particularly MRI films).
• What if the Neurologist does not know about ACC
  – Ask to meet again after the doctor has better familiarized him/herself with the issues
  – Ask for a referral to an expert in this area
• Don’t hesitate to schedule a follow-up visit to have the doctor answer more questions
• A one-time second opinion is not unwarranted even if you are happy with your child’s care.

**Possible Questions to ask:**
• What type of ACC is this?
• What are the associated CNS changes?
• What is your plan to get more information about our child’s condition?
• Is an EEG warranted?
• What “label” will you give to our child?
• When do you plan to get another MRI?
• When do you want to see our son/daughter next?

**Practical Considerations for any Doctor-Patient Relationship**
• Know all the members of your doctor’s team
  – *Doctor colleagues, nurses, staff*
• Know who to contact for what issue and how to reach them
• How do you get a hold of your doctor in emergencies and should you work out a contingency plan.
• Think of you and the doctor as a team working to help your child!

**ACC: Definitions**
• What type of ACC is this?
  – *Complete agenesis*
  – *Partial agenesis (hypogenesis)*
  – *Thin callosum (hypoplasia)*
• Remember: this is a radiographic diagnosis only, you will need more information to understand better the full nature of your child’s condition.

**The Normal Corpus Callosum**

**Planes of View: Head MRI**

**Adult Female, Normal CC**

**Complete Callosal Agenesis**

**Callosal Hypogenesis**

**Thin corpus callosum**

**Issues in MRI interpretation**
• What is the primary diagnosis?
  – *For example ACC can be part of a larger constellation of findings*
  – *This will be important for understanding range of outcomes, genetics and many other issues*
• What are the associated features (next talk)
  – *Hydrocephalus*
  – *Cyst*
• Is the interpretation accurate?
• When should you get a follow-up MRI
  – *Brain development, progressive condition, uncertain diagnosis*
MRI and other considerations

- Have the neurologist show you the films and point out the critical findings
- Get a copy of the films—both the films and a copy on disk (most images are initially stored electronically these days)
- Ask who read the films
  - General radiologist, neuroradiologist, pediatric neuroradiologist
- Ask whether a second opinion might be indicated (my general rule is always YES, no matter who has read them)

What caused the ACC?

- Genetic: known cause (Aicardi, Andermann, Mowat-Wilson, ARX etc)
- Likely genetic, but precise cause unknown
- Metabolic
- Injury/infection
- Unclear

My child has ACC: Now what do we do?

(part I-diagnostic evaluation)

- Other doctor visits (survey of issues)
  - Geneticist (MD, board certified in medical genetics)
  - Ophthalmologist
- Tests
  - Survey of other organs
    - Eyes, heart, kidneys, spinal cord
  - Genetic tests
  - Metabolic tests
  - Cause specific tests (for example stroke risk factors)

What will the geneticist do?

- Examine the body for other features that might provide a clue to the diagnosis
- Ask for pictures of the whole family
- Take an extensive family history
- Order a lot of tests
- Take a long time to sort things out—and maybe give a name to the condition.
- Help you understand recurrence risk!!
- Help you decide when to stop the evaluation

Genetic Tests

- High resolution chromosomes ($500)
- Subtelomeric Probes ($1200-$2000)
- Targeted testing
- Tests meant to survey the rest of the body
  - Echocardiogram, EKG, Renal ultrasound, X-rays of bones
- Research testing
  - Dr. Sherr’s group, for example

Chromosomal Analysis
Whole Genome Analysis (research based)

Example of Genomic Analysis

Possible Answers to Diagnostic Evaluation

- Identification of a specific genetic defect where gene is known
  - Can test other family members
  - Can perform prenatal testing or pre-implantation genetic diagnosis
  - No diagnosis based treatments for ACC, however
- Identification of genetic syndrome
  - Allows for understanding of range of outcomes and other associated issues
- Novel or uncommon genetic cause
  - Allows for recurrence risk counseling
- No definitive answer

What treatments are there?

- There are currently no "cures" for the underlying ACC (and other associated CNS changes)
  - The major structures of the brain were laid down at the end of the first and the second trimester of pregnancy
  - No evidence that other less conventional therapies work
    - Hyperbaric oxygen
    - Homeopathy
    - Stem cells--still too soon, issue of timing
  - Will early intervention cognitive therapies work?
    - Symptomatic Therapy

If your child has seizures, what do you want to know/ask?

- Seizures occur in 1/3 to 1/2 of ACC individuals
- Seizures are not caused by the ACC per-se, but by the associated miswiring of the brain
  - (seizures used to be controlled by cutting the corpus callosum to severe connections between the two cerebral hemispheres)
- Most patients have their seizures moderately to well controlled by medicine and 'seizure free' should be the initial goal

Seizure management-continued

- If you suspect seizures, how is the evaluation done?
  - Home video of seizure episodes, plus your description
  - EEG
  - Video EEG
- Treatments
  - Medicines (goal is to use only one)
  - Ketogenic Diet (only short-term solution)
  - Vagal Nerve stimulator
  - Balance for all between benefits and side-effects

Other Seizure Issues

- Have a plan in place for seizures that won’t stop
  - Additional medicines
School plan
How to get in touch with your Neurologist quickly
• Discuss safety and lifestyle issues
• When can your child come off medicine?
  – Understand natural history of seizures
  – Sooner controlled correlates with better outcome

Resources
• Most doctors don’t know this very well (myself included)
• Ask your doctor for referral to local agencies
• Ask your doctor to write in the visit letter the specific Symptomatic condition
  – Autism, cerebral palsy, etc
  – ACC diagnosis unlikely to bring services to your child
• Good place to start: www.govbenefits.gov
• For early intervention programs listing: www.nectac.org/contact/ptccoord.asp
• For Parent groups: www.taalliance.org/centers/index.htm

How is my child going to do?
• Most important question, least well understood for ACC
• Past performance is the best gauge of the future for individuals with ACC
• Landmarks
  – Walking, talking, mainstream classroom, living independently as an adult (the last two are the most difficult to predict)

Summary
• Visit a child neurologist in your community who is familiar with ACC
• Make this doctor your ally
• Keep copies of all your child’s records
• Get MRI’s at regular intervals until your child is grown and remains clinically stable
• Ask questions and write them down as they come up
• Pursue diagnostic evaluations, revisit this every one-to-two years to keep pace with discoveries
• Don’t hesitate to ask for a second opinion and/or consult with other doctors

Future Directions
• Genetic Research
  – Targeted analysis
  – Genome-wide scanning
• Imaging improvements
  – Prenatal MRI
  – Functional MRI
  – Diffusion tensor imaging
• Tailored therapy
  - Earlier intervention