

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 sever 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