CADASIL: Diagnosis and Testing

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Question

- How do you get diagnosed? I cannot find a doctor that seems to exhibit an interest. Can you recommend someone in the Colorado Springs/Denver area. --Lisa
General rules

- Each individual’s situation is different, so these are broad guidelines
Diagnosis

- Clinical assessment
- MRI
  - Genetic testing**
  - Skin biopsy*
Clinical assessment

- Your medical story
- Physical examination
- Family history
MRI

- White matter changes
- Some specific changes in CADASIL, but they are not absolute
Genetic testing or skin biopsy

- Should be obtained if suspicion is high for CADASIL
Genetic testing
(Drs. Kolodny and Herve)

- **NOTCH3** gene sequencing
  - Need to sequence the entire gene, unless another member of the family has CADASIL mutation
  - Athena
  - See Billie’s website
Skin biopsy

- Very good alternative to genetic testing
Personal decisions:
To test or not to test

- Personal and serious decision
- Refer to Sonia Reyes’ talk (before lunch)
Personal decisions: Why to NOT test

- “I just don’t want to know.”
- Possibility for increased stress
Personal decisions: Why to test

- Provides a definite diagnosis
  - Reduces additional tests
  - Some medications contraindicated
- Planning for the future
- Clinical trial eligibility
Summary: Diagnosis

- Clinical assessment
- MRI
  - Genetic testing*
  - Skin biopsy*
After diagnosis → testing
Testing: common studies

- Serial examinations (Neurologist)
- Neuropsychological examinations (Neuropsychologist)
- MRI (Radiologist)
Serial examinations

- The interval history
  - Mood disturbances?
  - Agitation? (Refer to Dr. Galvin’s remarks)

- Serial physicals
  - Brief mental status examination
  - Neurological examination
Neuropsychological examination

- Longer than your doctor’s visit
- More detailed assessment
- Can define strengths and weaknesses
- Can detect depression, apathy
- Can suggest coping strategies
- Not always necessary
MRI

- Were there any new strokes?
- How fast are things changing?
- Not always necessary