Genetics of TTR Amyloidosis

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Everyone has two copies of the TTR gene
TTR Amyloidosis follows an “autosomal dominant” inheritance pattern
- Cardiomyopathy
- Carpal Tunnel Syndrome
- Neuropathy

New diagnosis of TTR Amyloidosis
Genetic Testing: Why do we do it?

- Confirm whether the amyloidosis in your family is genetic vs. wildtype
- Impacts treatment and eligibility to clinical trials
- Necessary for testing of family members
- Can provide information about what to expect medically
Different mutations have different names
TTR GENE

Val30Met  Thr60Ala  Tyr114Cys

Val20Ile  Ile84Ser  Val122Ile
Mutations can cause different common symptoms

- **Val122Ile** - Heart issues
- **Asp18Glu** - Peripheral neuropathy
- **Gly47Ala** - Peripheral and autonomic neuropathy
- **Ile84Ser** - Heart and eye issues, carpal tunnel syndrome
So what does this mean for my family?

- Heart issues
- Carpal Tunnel Syndrome
- Neuropathy

New diagnosis of TTR Amyloidosis
How Do I Share This Information with my Loved Ones?

- Open communication!
- Family letter
- Other resources
  - Amyloid Support Group website!
- Identify the family communicator
- You know your family best!
So what does this mean for my family?

Dad

Mom

Ben

Emily

Mike

Sarah
I don't have symptoms of TTR amyloidosis, Should I be tested?
No way! I do NOT want to be tested for TTR Amyloidosis!

Mike
Should I Get Tested?

• Genetic testing is a personal choice
• Help make life choices  
  o Lifestyle choices  
  o Reproductive decisions
• Relieve anxiety of uncertainty
• Know for the sake of children
• Allow for earlier diagnosis and clinical monitoring

• Genetic testing is not a crystal ball!
Sarah

How will I react if I am positive?

How will I use this information?

How will I feel if I am not at risk?

Am I able to handle the impact of the positive result?

Is now the right time?
What about my insurance?

• Insurance may cover the cost of the testing

• Genetic Information Non-Discrimination Act
  o Protects most individuals from discrimination for:
    • Health Insurance
    • Employability
  o Does not apply to:
    • Life Insurance
    • Disability Insurance
    • Long-term Care Insurance
Testing Logistics

• Testing usually performed through blood draw

• Analysis of known familial mutation in TTR gene
  o Or other gene causative of amyloidosis in some cases!

• Two to Three weeks later…
Okay… results are in.

- **Positive Result**
  - Mutation identified
  - Inherited mutation
  - Seek care with physician and medical team with experience in TTR Amyloidosis
  - Continue with regular evaluations

  [Image of Sarah]

- **Negative Result**
  - No mutation identified
  - Did not inherit mutation
  - No further evaluations needed

  [Image of Ben and Emily]
Who else is at risk?

• All first degree relatives of an individual with familial amyloidosis have a 50% chance of also carrying the gene mutation
  o Children
  o Siblings
  o Parents

• Symptoms are variable! Even in the same family.
  o Different age of onset
  o Severity of symptoms
  o Types of symptoms
How Do I Get Tested?

• Meet with genetics professional
• www.findageneticcounselor.com
Thank you!

Questions?

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