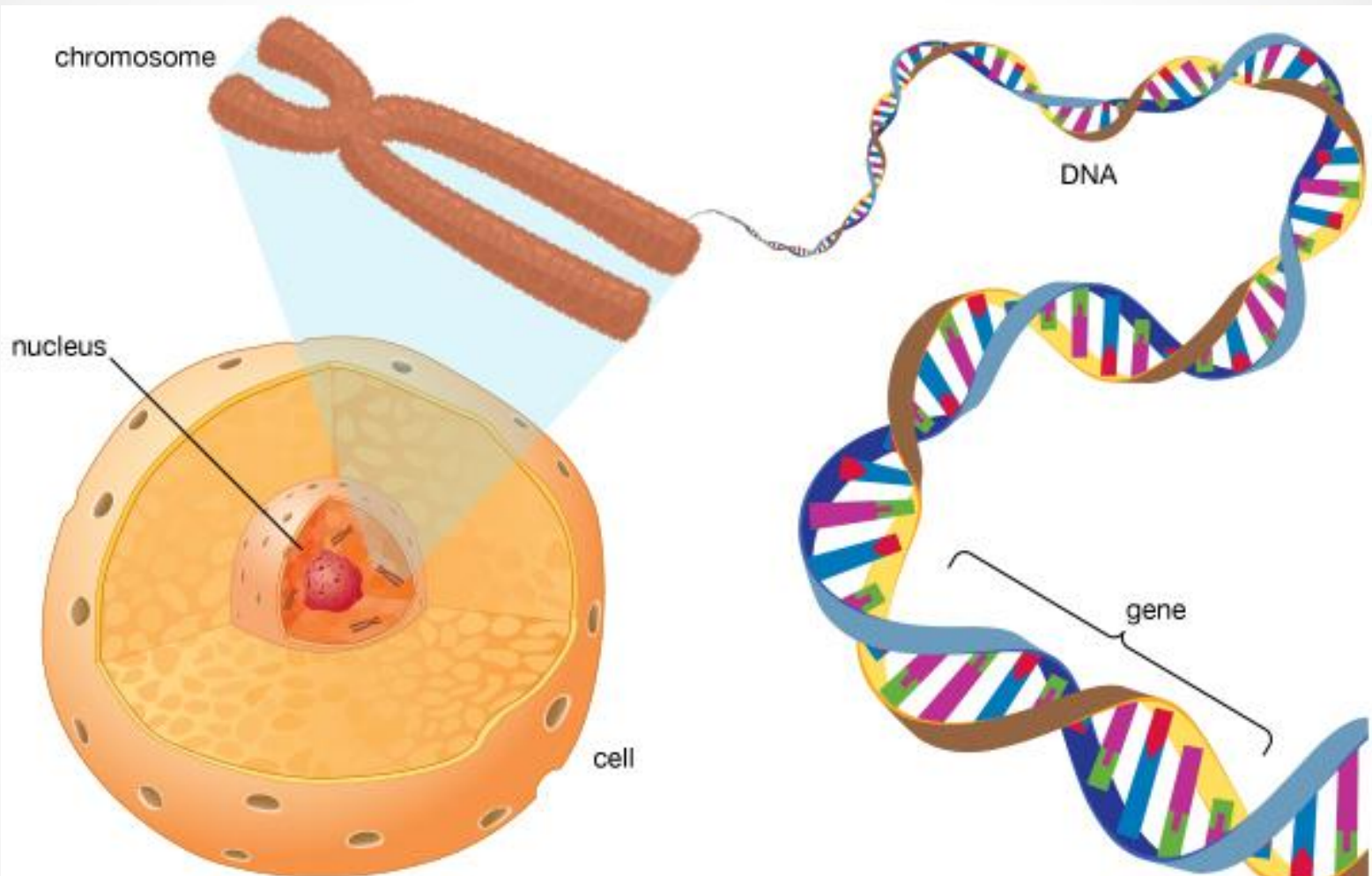
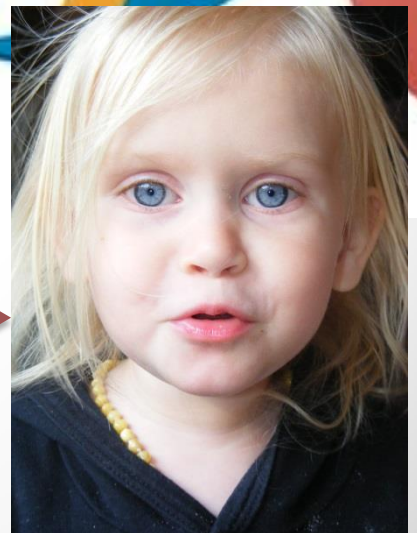
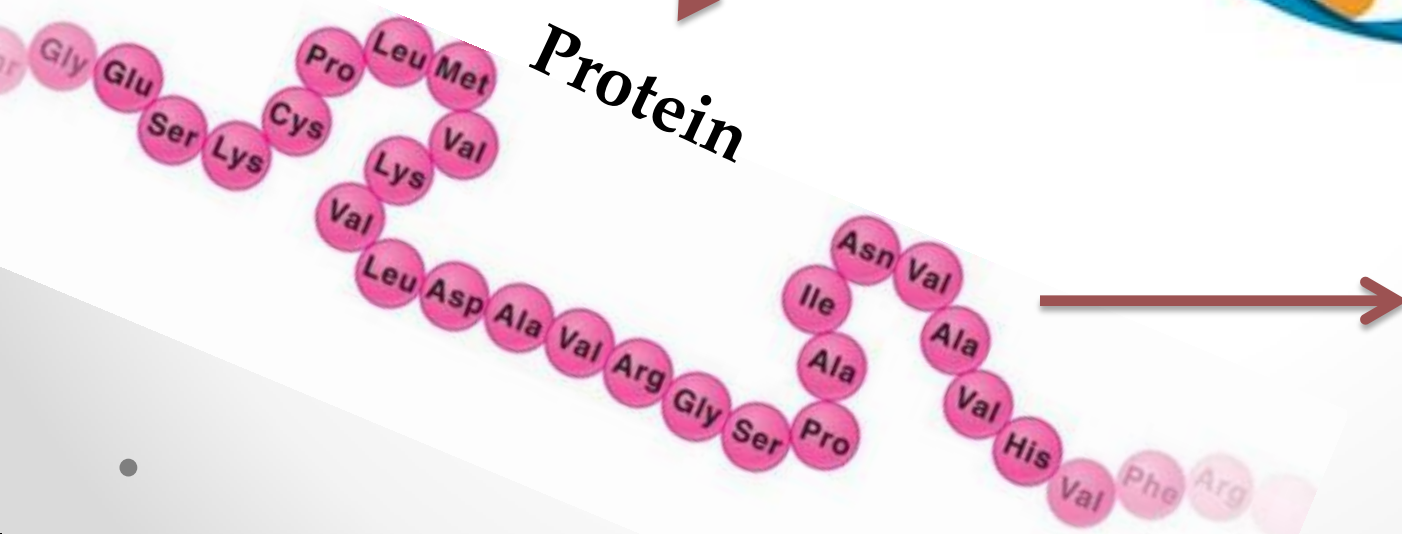
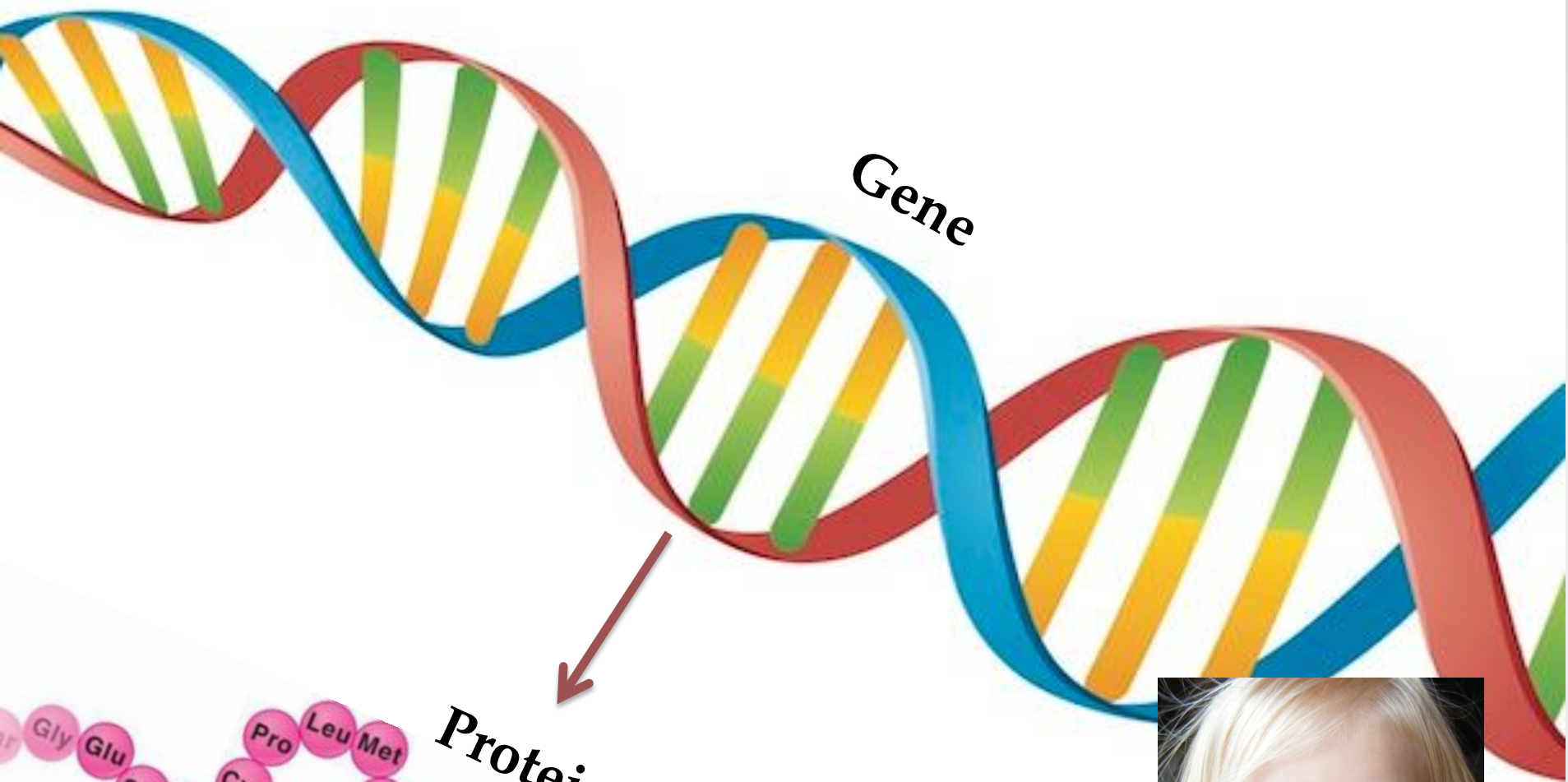
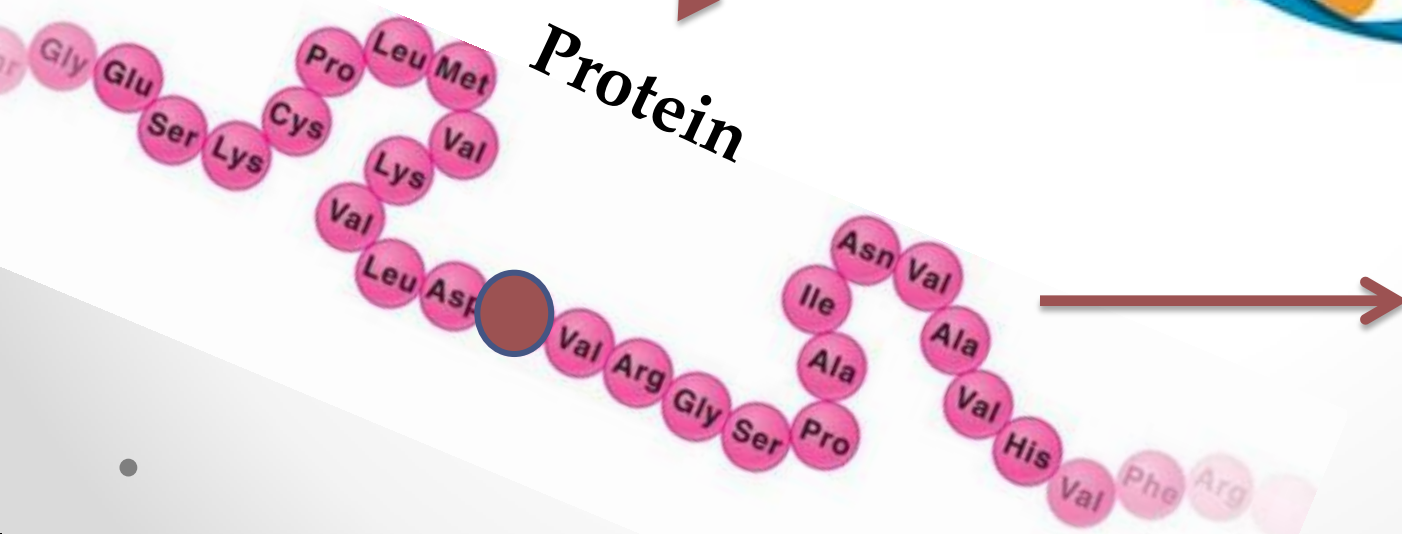
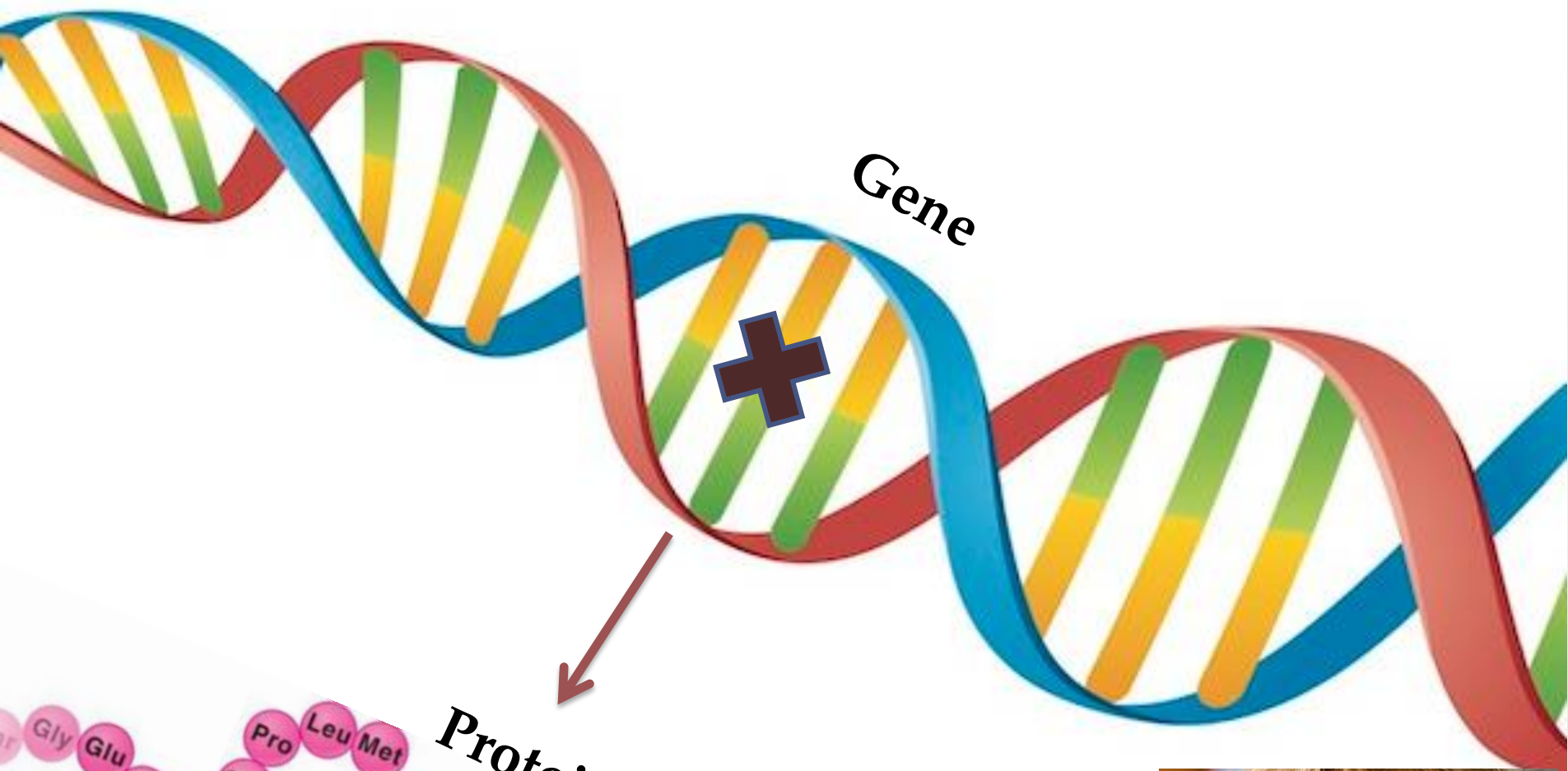


Genetics of TTR Amyloidosis

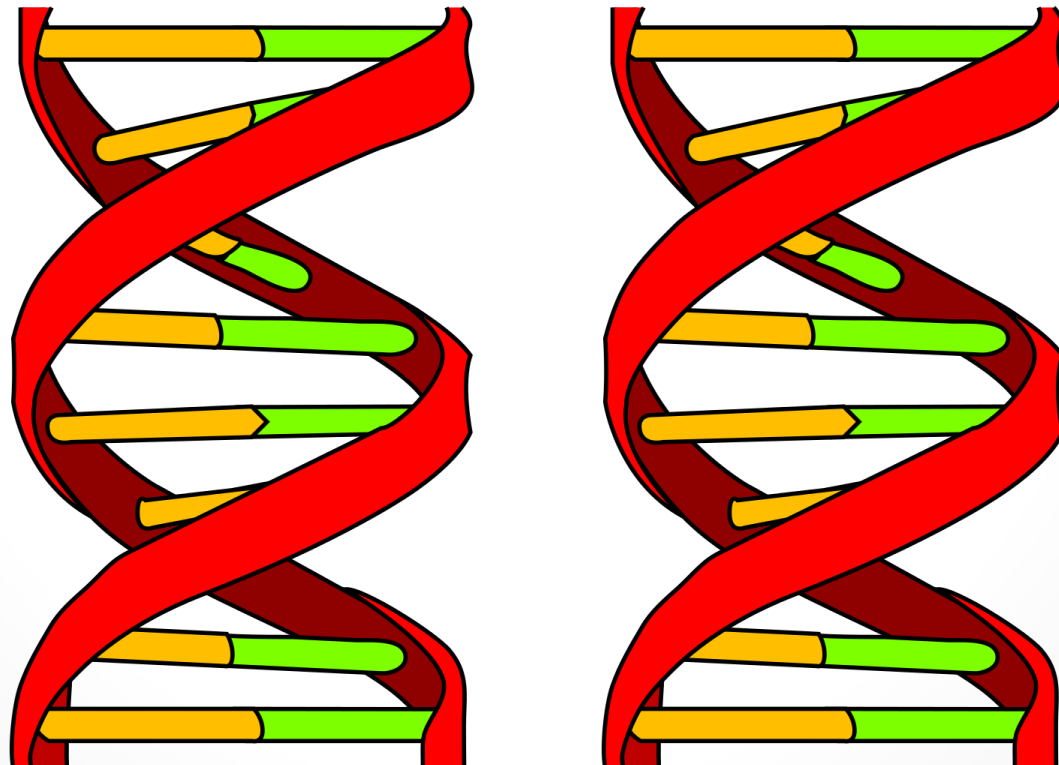
Katie Agre, MS, LCGC
Licensed Genetic Counselor
Mayo Clinic



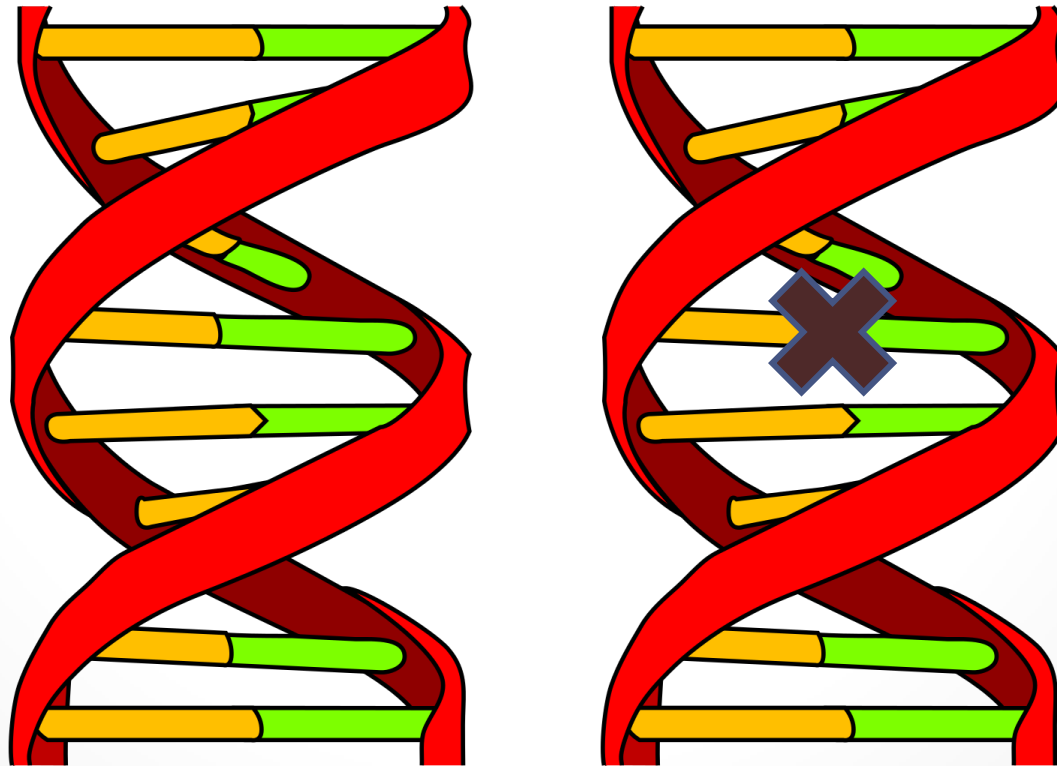




Everyone has two copies
of the TTR gene



TTR Amyloidosis follows an
“autosomal dominant”
inheritance pattern



- Cardiomyopathy
- Carpal Tunnel Syndrome
- Neuropathy

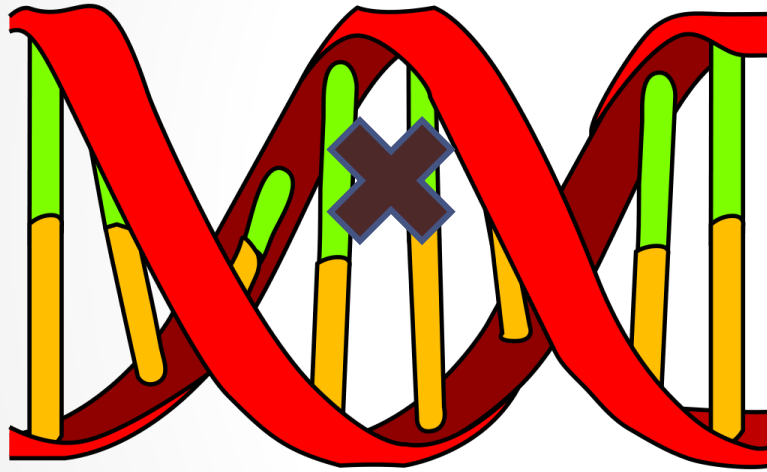
New diagnosis of
TTR Amyloidosis



Genetic Testing: Why do we do it?

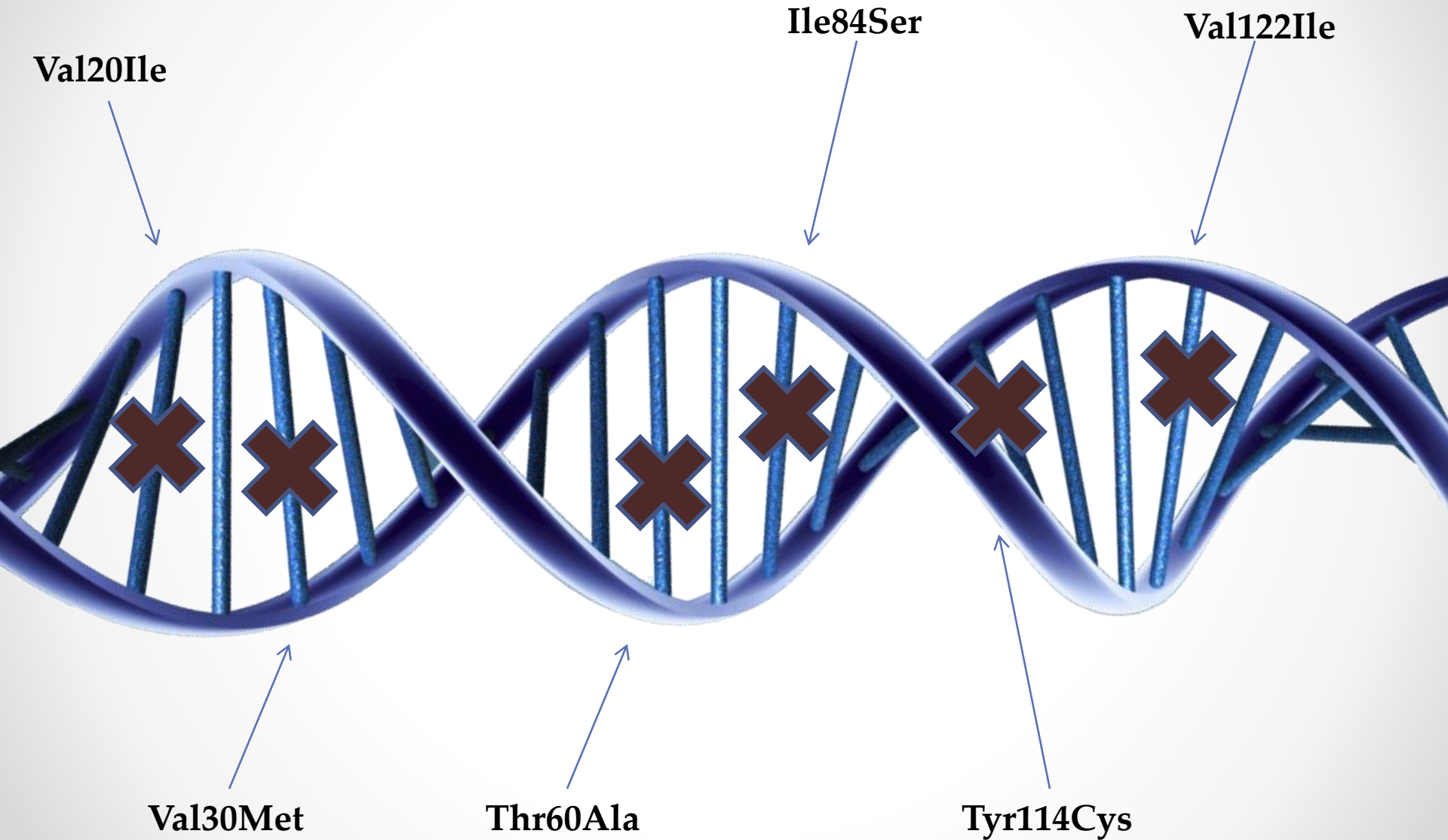
- Confirm whether the amyloidosis in your family is genetic vs. wildtype
- Impacts treatment and eligibility to clinical trials
- Necessary for testing of family members
- Can provide information about what to expect medically





Different
mutations
have different
names

TTR GENE



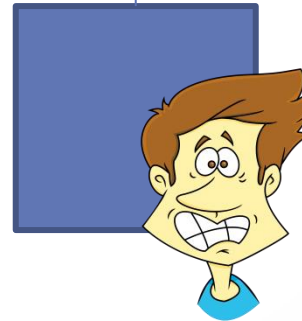
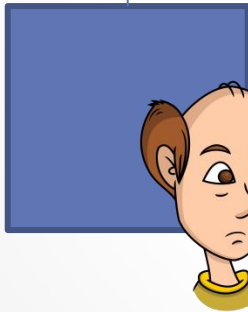
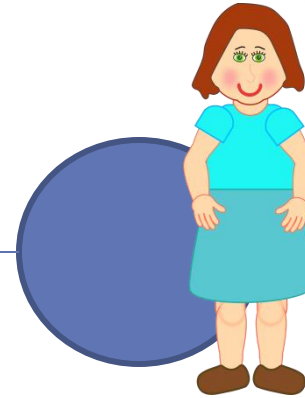
Mutations can cause different common symptoms

- **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

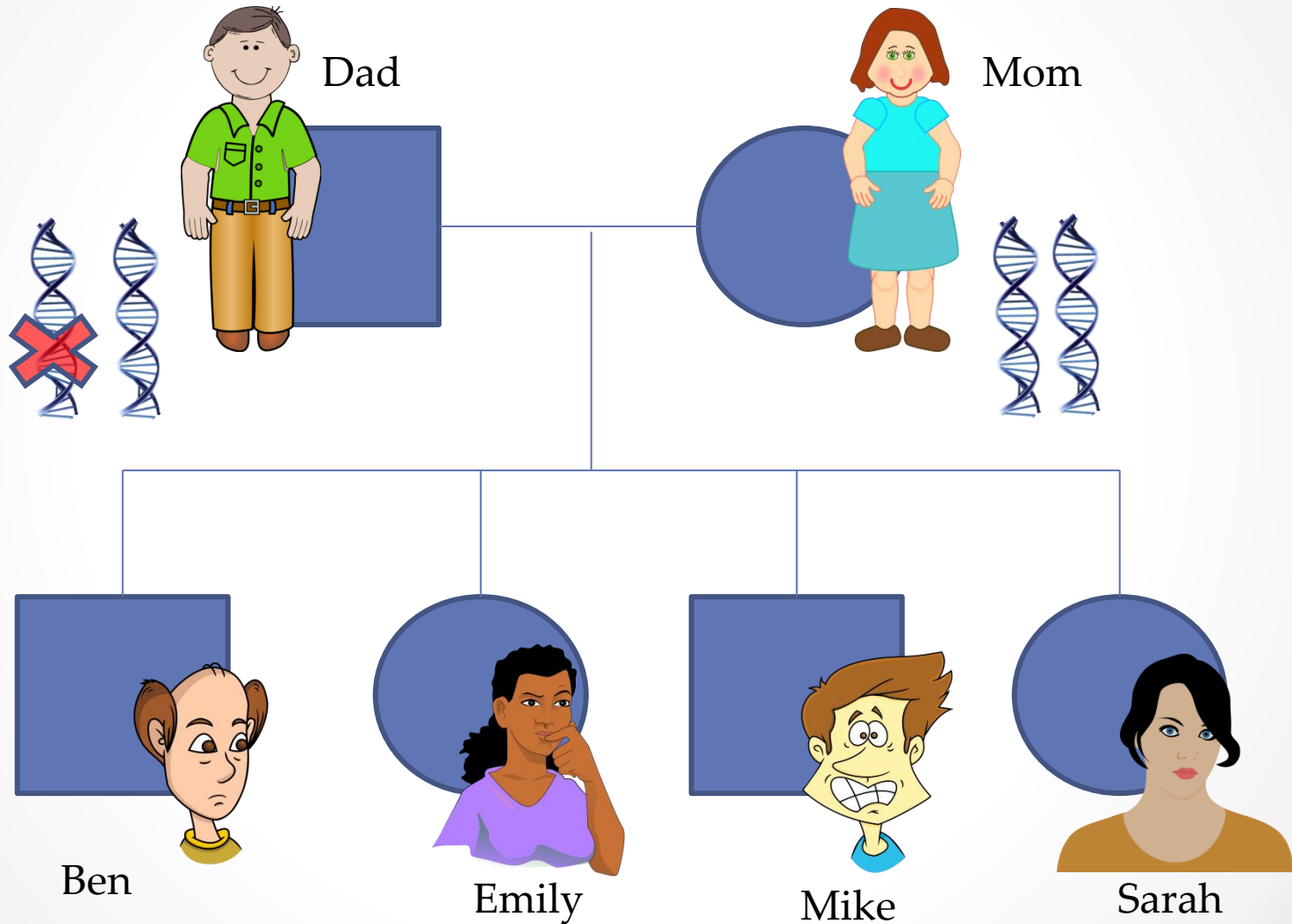


How Do I Share This Information with my Loved Ones?

- 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?



I don't have symptoms of TTR amyloidosis,
Should I be tested?



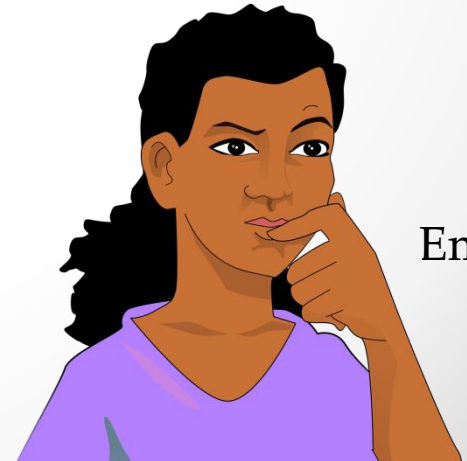
Ben



Sarah



Emily



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
 - Lifestyle choices
 - 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!



How will I feel if I am not at risk

How will I react if I am positive?

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

How will I use this information?

Is now the right time?



Sarah

What about my insurance?

- Insurance may cover the cost of the testing
- Genetic Information Non-Discrimination Act
 - Protects most individuals from discrimination for:
 - Health Insurance
 - Employability
 - 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
 - Or other gene causative of amyloidosis in some cases!
- Two to Three weeks later...

Okay... results are in.

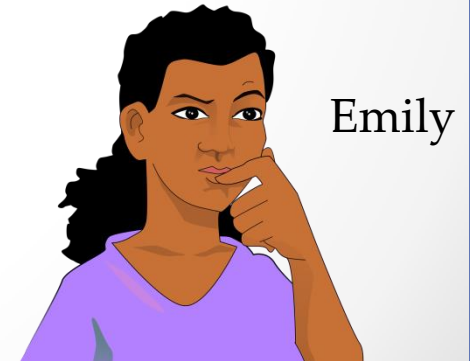
- **Positive Result**

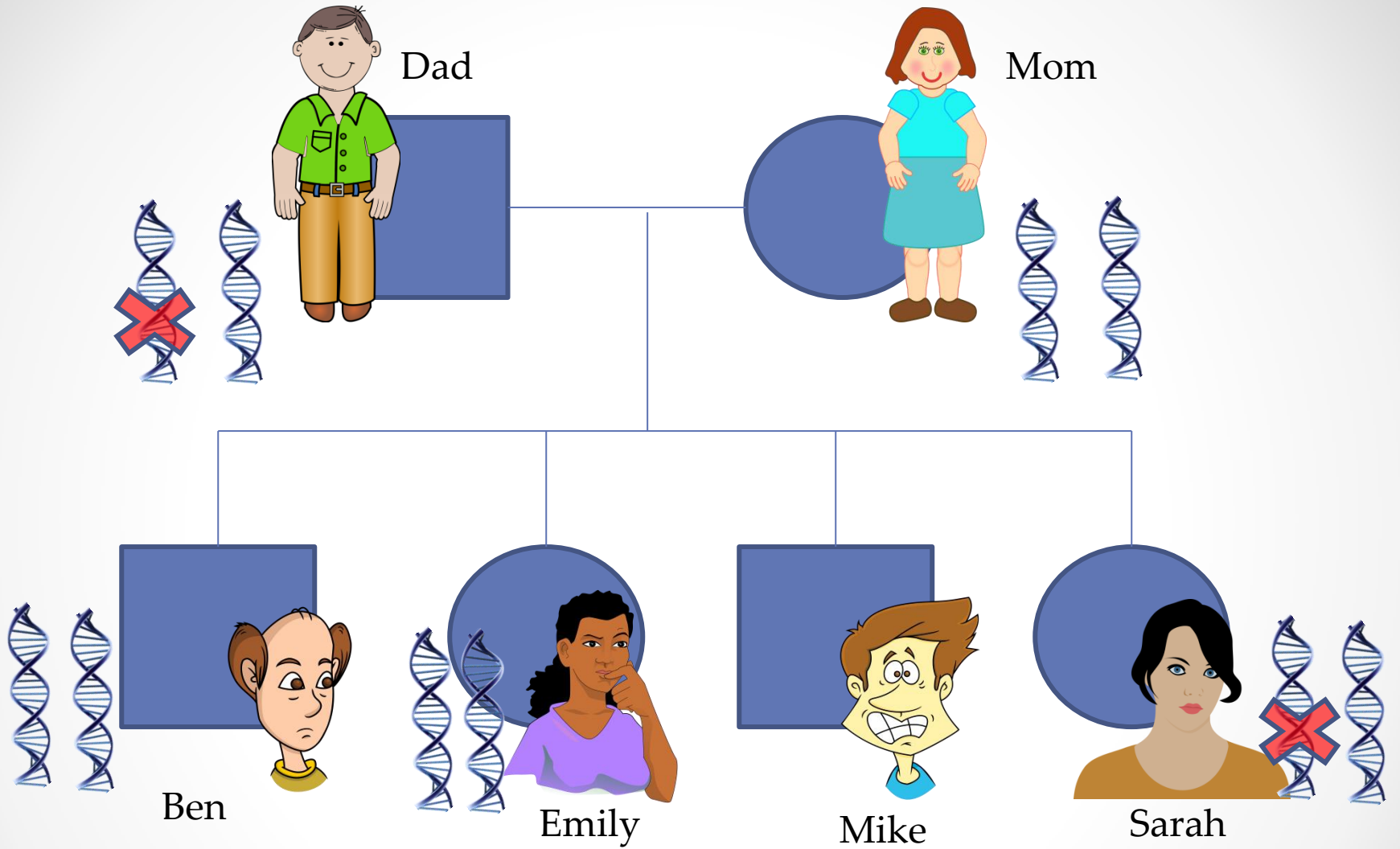
- Mutation identified
- Inherited mutation
- Seek care with physician and medical team with experience in TTR Amyloidosis
- Continue with regular evaluations



- **Negative Result**

- No mutation identified
- Did not inherit mutation
- No further evaluations needed





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
 - Children
 - Siblings
 - Parents
- Symptoms are variable! Even in the same family.
 - Different age of onset
 - Severity of symptoms
 - Types of symptoms

How Do I Get Tested?

- Meet with genetics professional
- www.findageneticcounselor.com

Thank you!

Questions?

agre.katherine@mayo.edu