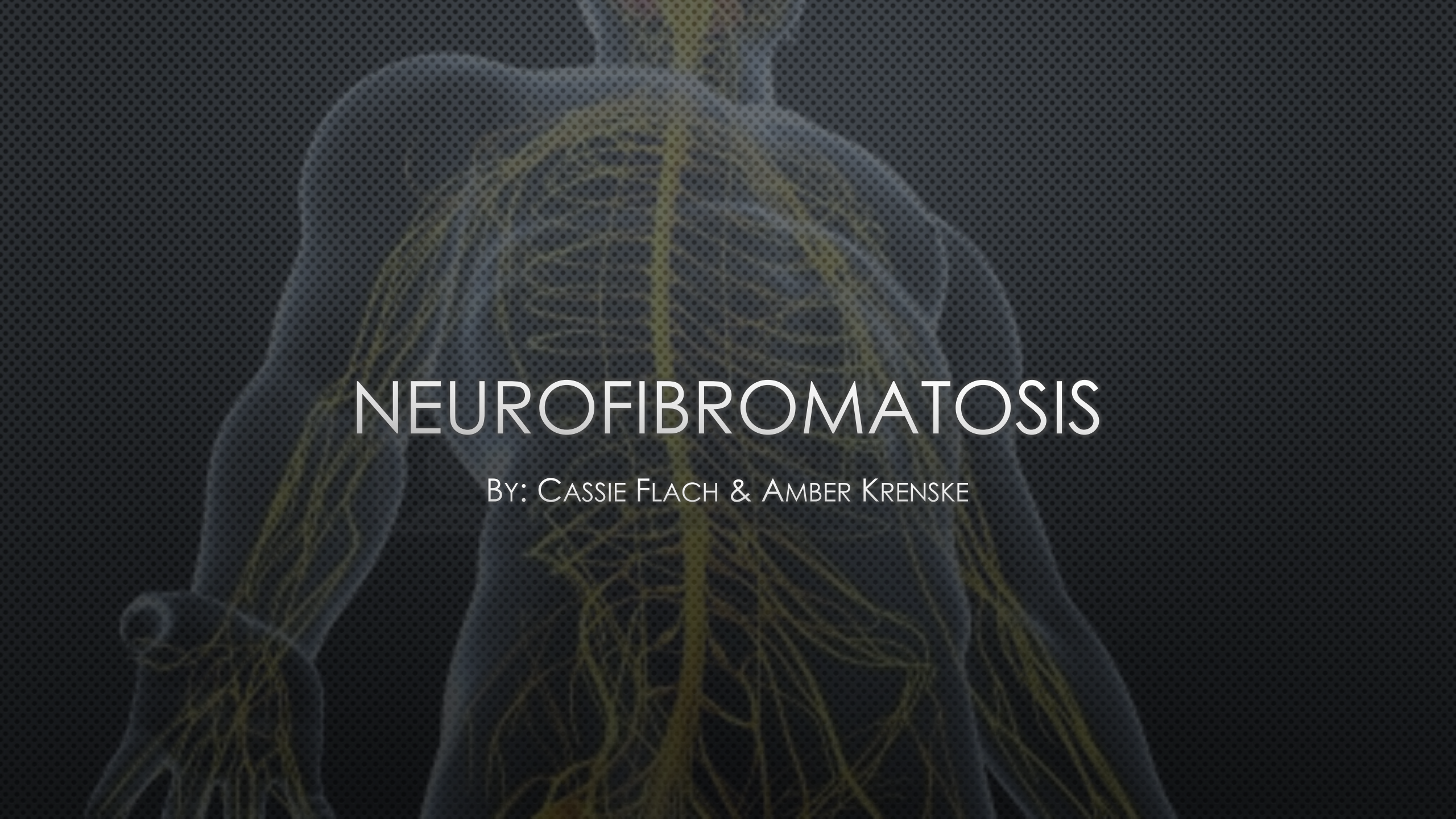
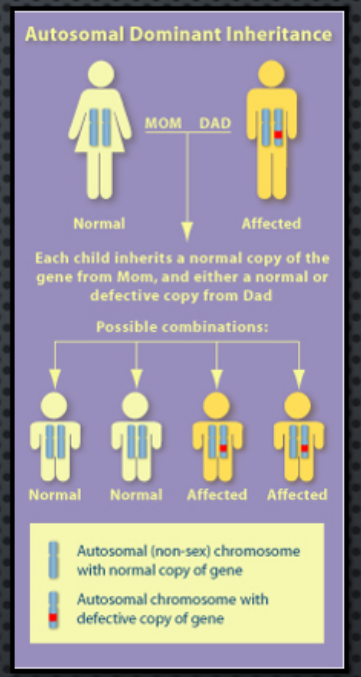


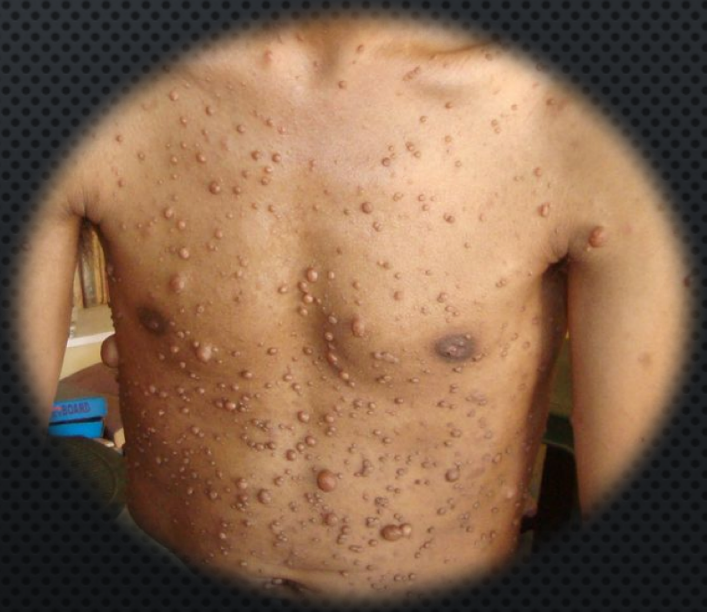
NEUROFIBROMATOSIS

By: Cassie Flach & Amber Krenske

WHAT IS IT?

- Progressive disorder
- Autosomal Dominant
  - Inherited from an affected parent OR spontaneous mutation
- Mutation in the NF1 Gene
  - Neurofibromin – tumor suppressor
  - Tumor growth mainly on nerves of skin, brain, and spinal cord
- One of the most common genetic disorders in the US
  - 1 in 3000-4000 people worldwide (NF1)
- Variable – mild vs. Severe cases
- No Known Cure





TYPES

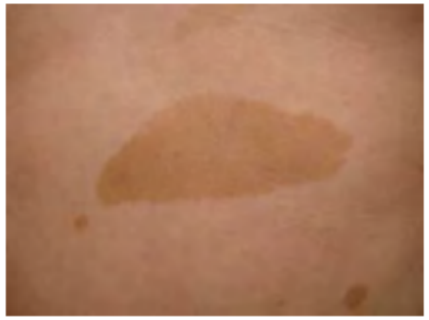
- **Neurofibromatosis 1 – most common**
- Neurofibromatosis 2
  - Affects auditory & vestibular nerves
    - Problems with balance & walking, tinnitus, dizziness, headaches, hearing loss, numbness/pain
  - Typically presents during adolescence/early adulthood
- Schwannomatosis
  - "Related" Disorder
  - Presents age>30

Diagnosis

- **Signs/Symptoms**
  - Growth of neurofibromas
  - Changes in skin pigmentation
    - Café-au-lait spots
    - Freckles in underarm and groin
  - Lisch nodules
  - Short stature
  - Macrocephaly
  - Scoliosis
  - Bowing of tibia

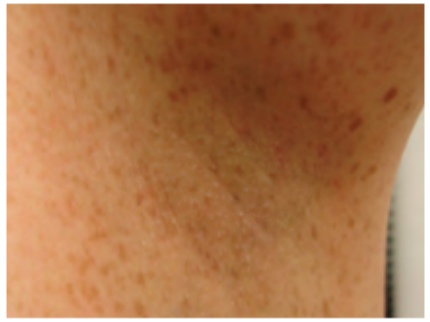
- **Clinical Diagnosis**
  - Physical examination + family history
  - Genetic testing
  - Imaging
  - Biopsy of tumor tissue
  - Eye and ear exams

Two signs/symptoms required for diagnosis



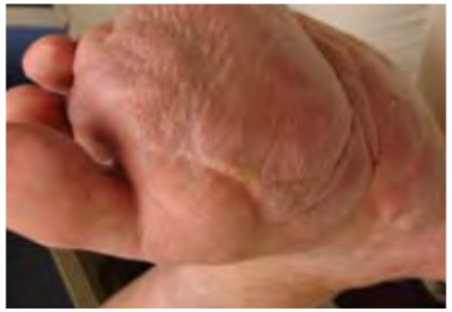
Café-au-lait Macules

Flat, darkly pigmented spots of birthmarks on the skin, typically present at birth.



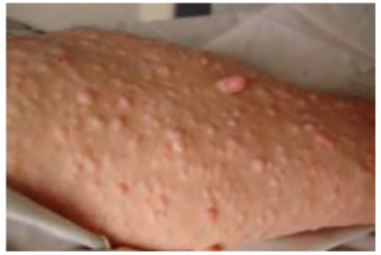
Skinfold Freckling

These freckles are commonly seen in areas of the body not exposed to the sun, like the armpits and groin. They also may be found under the neck or under the breasts in women.



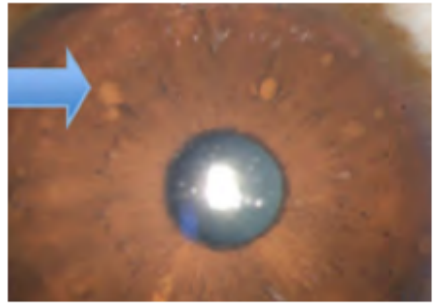
Plexiform Neurofibromas

25 to 30 percent of individuals with NF1 will develop a larger, more diffuse type of neurofibroma, termed a plexiform neurofibroma, which can grow to large proportions and affect adjacent structures and organs. Rarely, these tumors can become malignant.



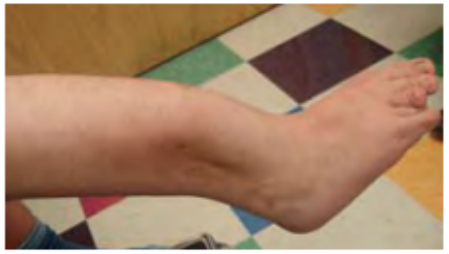
Neurofibromas

Benign growths which typically develop on or just underneath the skin but may also occur within the body. These are seen in nearly all adults with NF1. These tumors are not contagious. NF is progressive, and the majority of people will experience increases in tumor numbers and size. Regular evaluations in a coordinated care clinic specializing in NF1 is necessary to identify and address potential problems early.



Lisch Nodules

These nodules are benign pigmented growths on the iris (the colored portion of the eye). They are usually found in both eyes but do not interfere with vision.

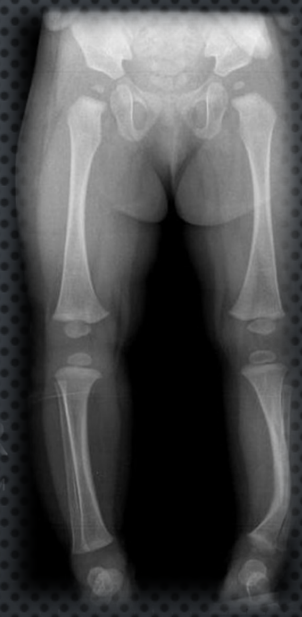


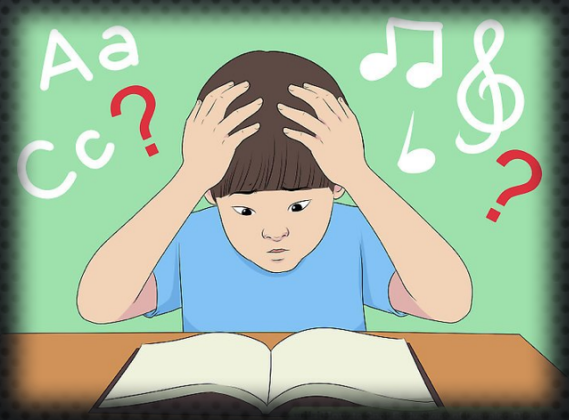
Bone Abnormalities

Children with NF1 are prone to the development of bone deformities involving the lower leg, forearm and eye socket. Deformities of the lower leg and forearms can cause a bowing of these bones and lead to repeated fractures. Bone problems involving the eye socket can affect the way the eye sits in the socket. All of these deformities are typically noticed early in childhood and are treated promptly.

MEDICAL COMPLICATIONS

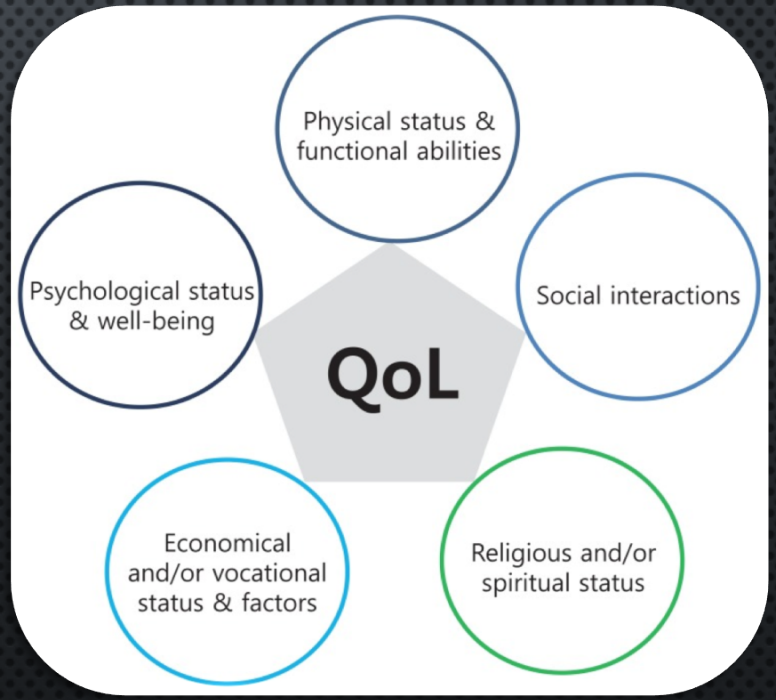
- **PREDISPOSED TO**
  - LEARNING, BEHAVIORAL, AND SOCIAL DIFFICULTIES (~66%)
  - BONE ABNORMALITIES (~25%)
  - SEIZURES AND EPILEPSY (~6-9%)
  - DEVELOPMENT OF OTHER CANCERS
  - HYPERTENSION
  - VISION LOSS
  - HEADACHES AND MIGRAINES
  - RENAL ARTERY STENOSIS





Quality of Life

- High quality of life with this diagnosis
- Normal intelligence
  - Learning disabilities/ADHD
- Increased risk of certain cancers
- Concerns about appearance may cause emotional distress
- Family counseling, genetic counseling, and support groups
- Life expectancy is less than average



APPLICATION TO PHYSICAL THERAPY PRACTICE

- Neurofibromatosis is variable with systemic involvement
  - Musculoskeletal: bony abnormalities, pain
  - Neuromuscular: motor proficiency impairments
  - Cardiovascular: monitor blood pressure
- Best physical therapy practice has yet to be determined
  - A series of case reports suggest benefits from stretching, strength training, postural training, and palliative care
  - Standard of care – Early diagnosis and multidisciplinary team

IN THE LITERATURE

- 70 children with NF1
  - Movement assessment battery for children (MAB-C)
  - Intelligence, emotional and behavioral problems as rated by parents
  - 61% of the children had severe motor problems and another 17% were borderline
- Motor problems are broad; similar to DCD
  - Independent of intelligence or age
- **We can assess and treat motor coordination impairments with physical therapy!**

CAMERON'S COUSINS' STORY

RESOURCES

- https://ghr.nlm.nih.gov/condition/neurofibromatosis-type-1
- https://www.nfnetwork.org/
- https://www.nfnetwork.org/data/uploads/nf1-educational-materials/clinical-care-options-2016.pdf
- https://www.aans.org/Patients/Neurosurgical-Conditions-and-Treatments/Neurofibromatosis
- https://orthoinfo.aaos.org/en/diseases--conditions/neurofibromatosis
- https://ghr.nlm.nih.gov/condition/neurofibromatosis-type-2
- https://www.physio-pedia.com/Neurofibromatosis_Type_I
- Rietman AB, Oostenbrink R, Bongers S, et al. Motor problems in children with neurofibromatosis type 1. *J Neurodev Disord*. 2017;9:19. Published 2017 May 19. doi:10.1186/s11689-017-9198-5
- https://escholarship.org/uc/item/29s5592h