

Neurofibromatosis Type 1

Uncommon but Important
Health Considerations in adolescence

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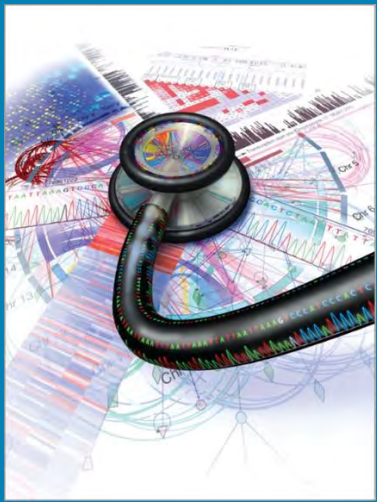


Disclosures and Conflicts of Interest



I consult for PPD, a company that oversees clinical trial safety monitoring. There is no relevant financial relationship to disclosure related to my presentation today

TKO



Topics for today's discussion

Uncommon things in NF1

Transition into Adult Health care

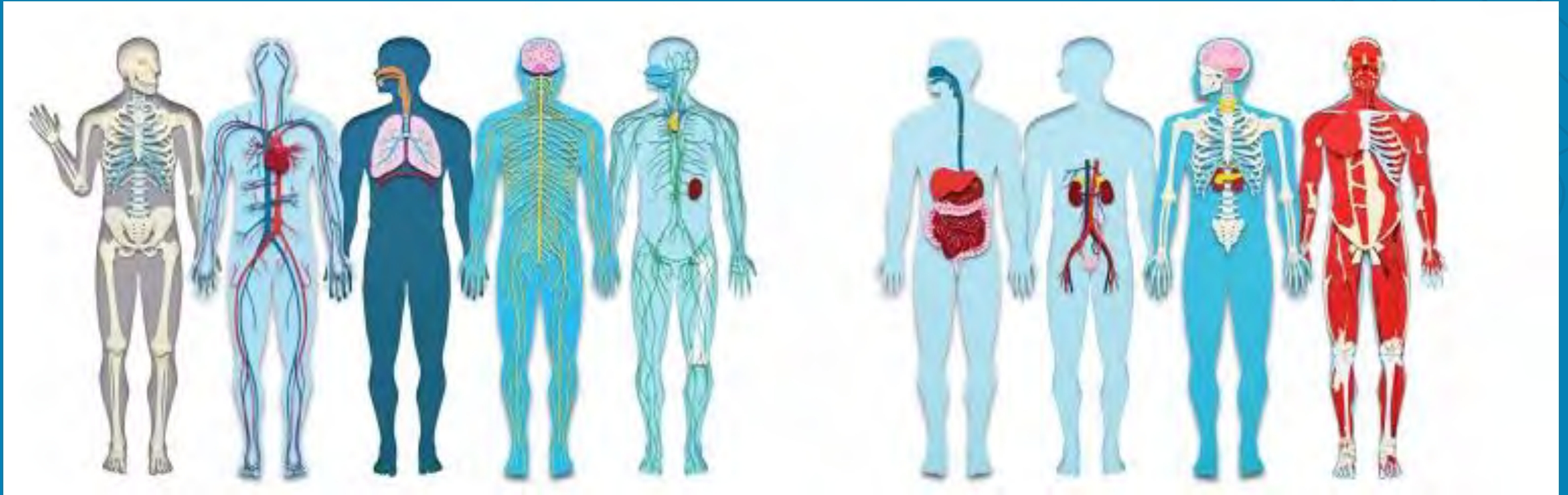


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Uncommon things in NF1



Dorling Kindersley: Owen Gildersleeve / Getty Images

Let's think about what systems can be involved



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Uncommon things in NF1

We won't be talking about:

- Neurofibromas
- Optic Gliomas
- Café au lait macules

BUT if you have questions

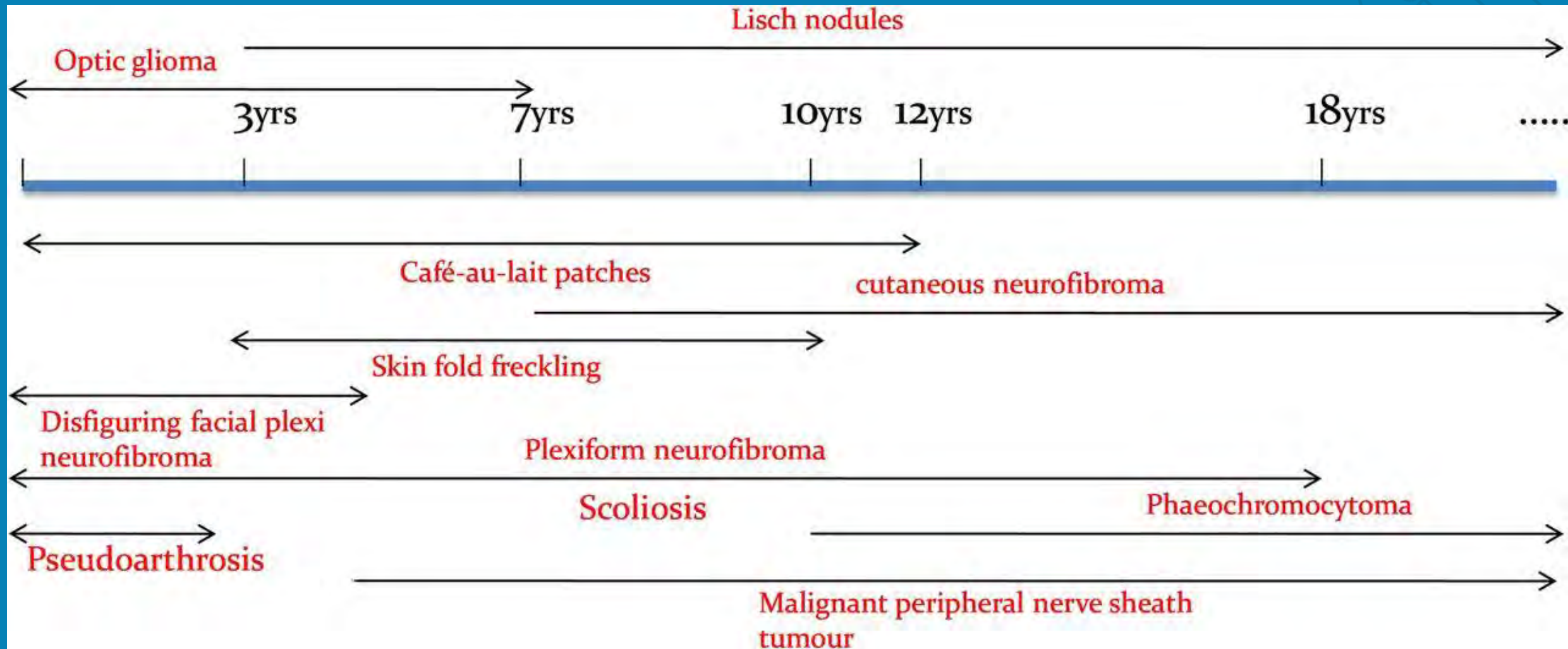
- feel free to ask about them when we have our Q&A



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Neurofibromatosis type 1 (NF1) time line.

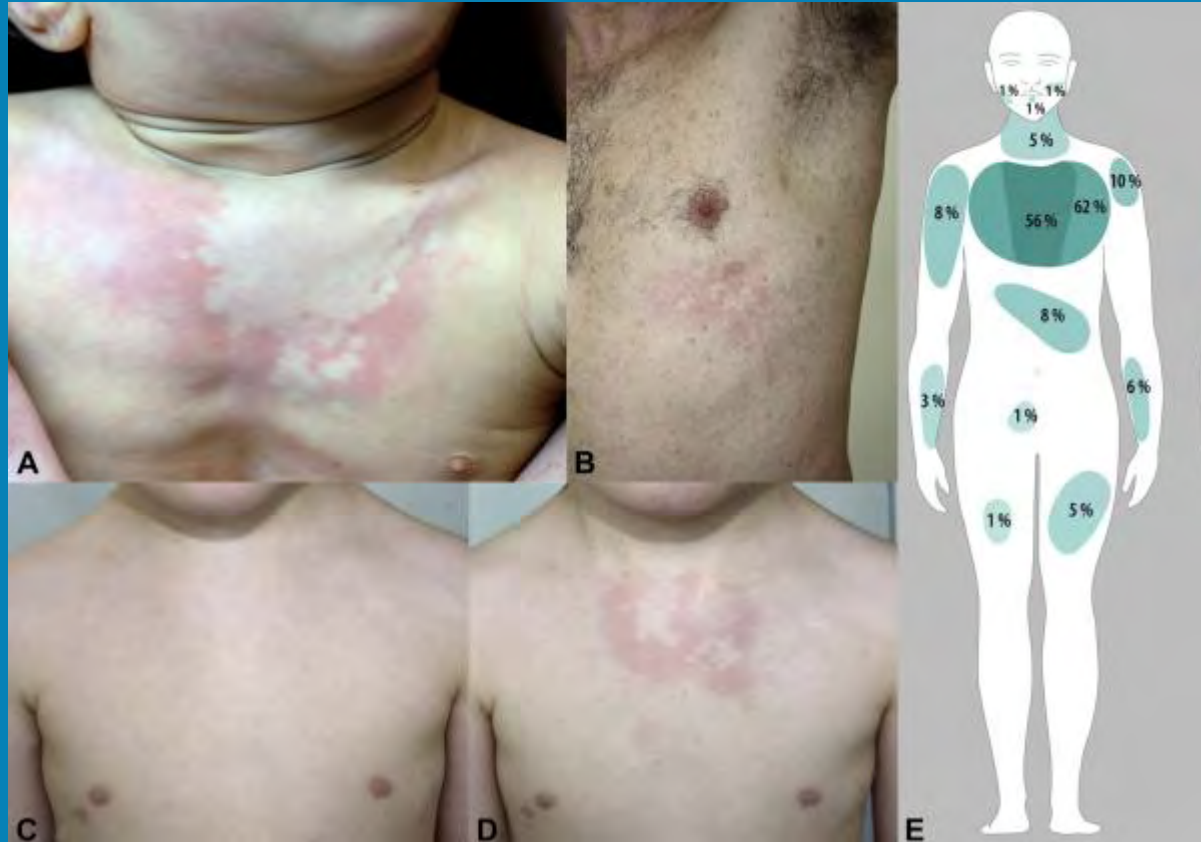
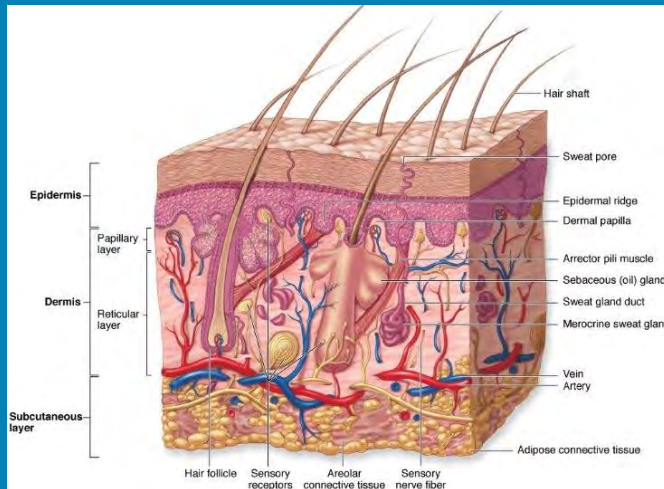


BM Dunning-Davies, and APJ Parker Arch Dis Child Educ Pract Ed 2016;101:102-111

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Uncommon things in NF1

Skin



<https://doi.org/10.1016/j.jaad.2013.06.039>

Nevus Anemicus

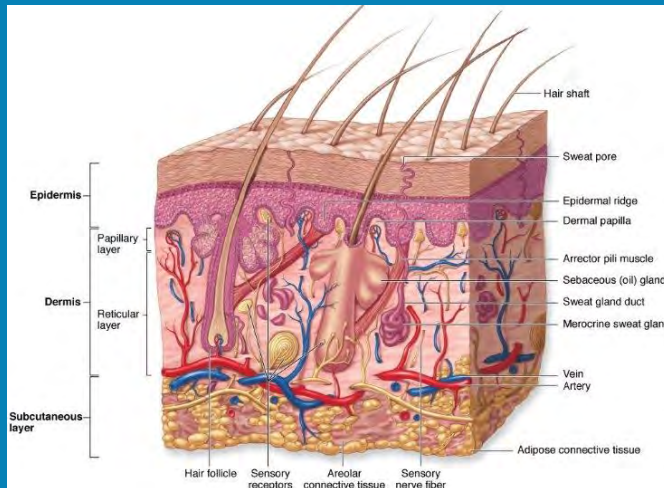


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Uncommon things in NF1

Skin



<http://medcomhk.com/hkdvb/pdf/2009v17n02-05.pdf>

Juvenile xanthogranuloma

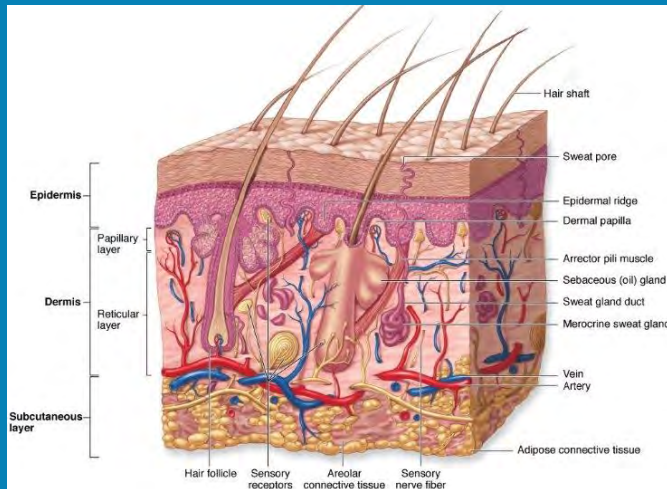


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Uncommon things in NF1

Skin



Pruritis = itchiness

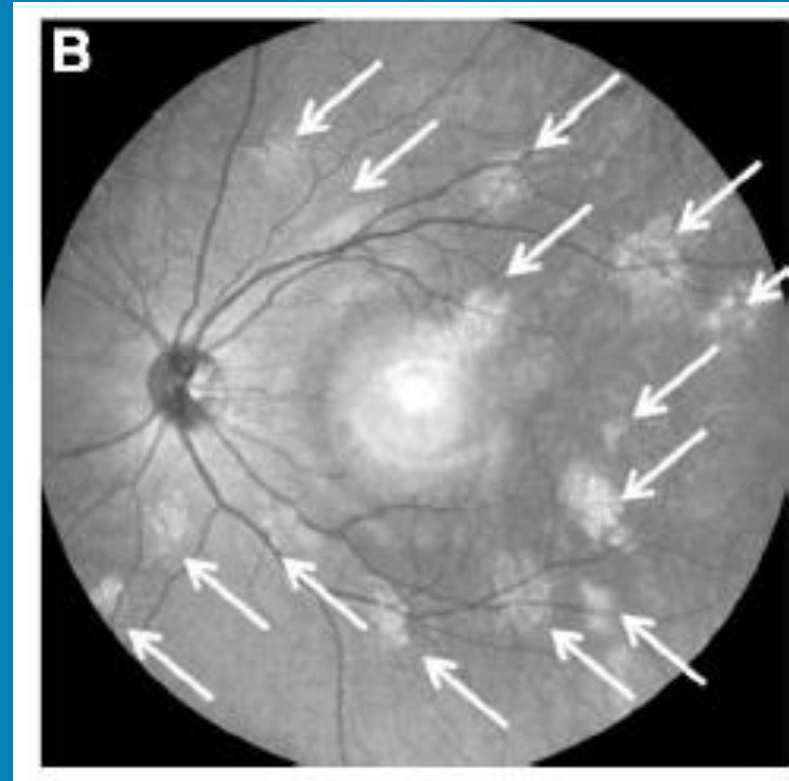


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Uncommon things in NF1

Eye



<https://doi.org/10.2147/OPHTH.S56327>

choroidal freckling

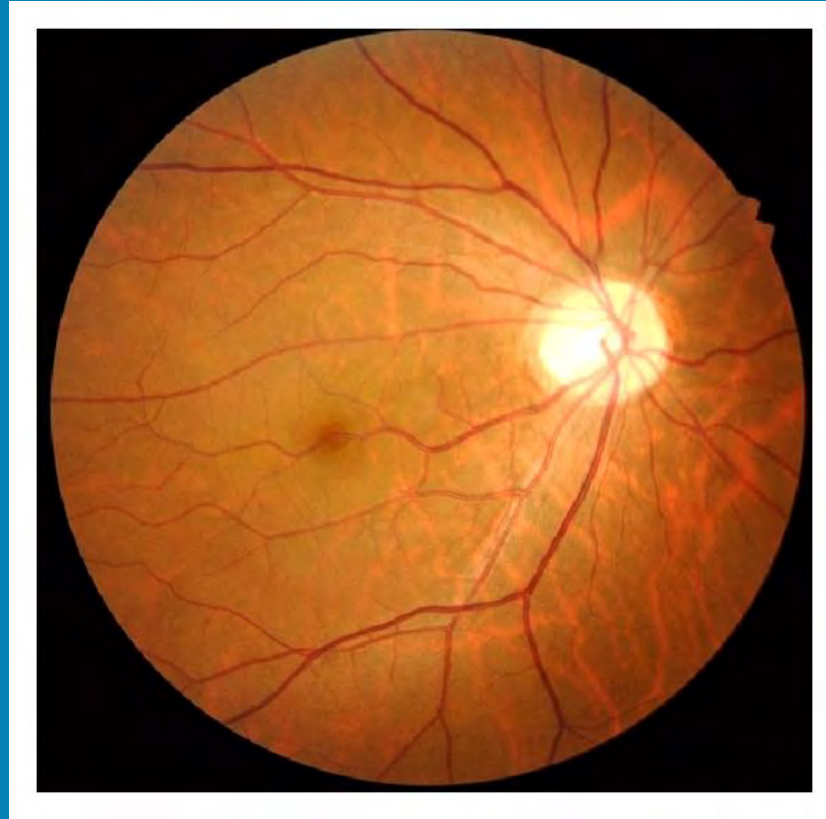
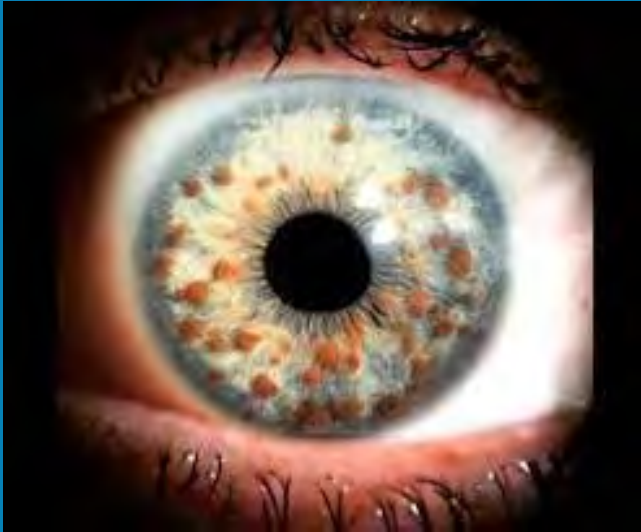


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Uncommon things in NF1

Eye



<https://doi.org/10.1155/2013/604191>

Retinal Macrovascular anomalies



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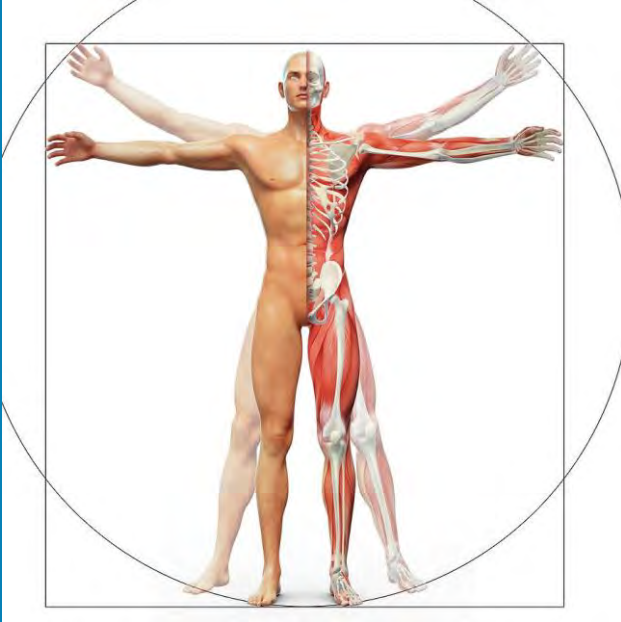


Uncommon things in NF1

Musculoskeletal system

BONES:

- lower mineralization = osteopenia / osteoporosis
 - lower Vit D levels
 - increased Parathyroid hormone
 - higher bone resorption
- Dysplasia of bones
 - Sphenoid wing dysplasia
 - Pseudoarthrosis
 - Vertebral dysplasia – severe early onset scoliosis “Dystrophic”
 - Dural Ectasia



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Uncommon things in NF1



Pseudoarthrosis



Sphenoid wing dysplasia



Dural Ectasia



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Uncommon things in NF1

Musculoskeletal system



Muscles:

- Reduced Strength
- Fatigue

Joints:

- Hypermobility
 - Hand cramping, flat feet, growing pains
- Instability
 - “rolling ankles” / strains and sprains



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Uncommon things in NF1

Learning:

Neurological System



- Intelligence is normal
 - 50-80% incidence of :
 - Learning difficulties
 - Visio-spatial
 - Behavioral problems
 - Attention and Social awareness
- More recent studies suggests higher incidence of Autism (as high as 30%)
- Polyneuropathy
 - Multiple nerve root tumors
- Seizures
- Abnormal sleep - difficulty falling asleep , sleep cycle dysregulation



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Uncommon things in NF1

Wellness:

Neurological System



- Anxiety

- Impacts how teenagers interact with their world
- Affects confidence

all impact how children and teenagers/adults cope with life experiences

Overall Well-being Boosters

- Exercise
- Sleep
- Nutritious eating
- Social interaction



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Uncommon things in NF1



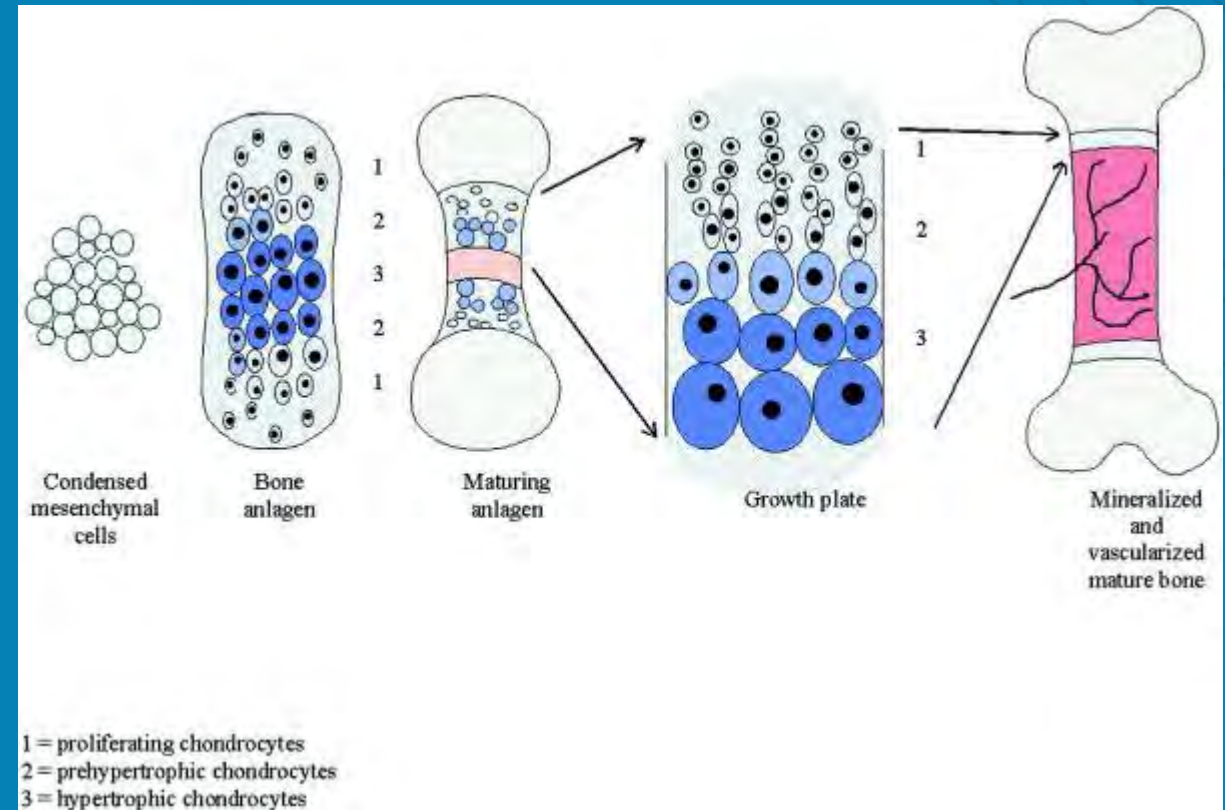
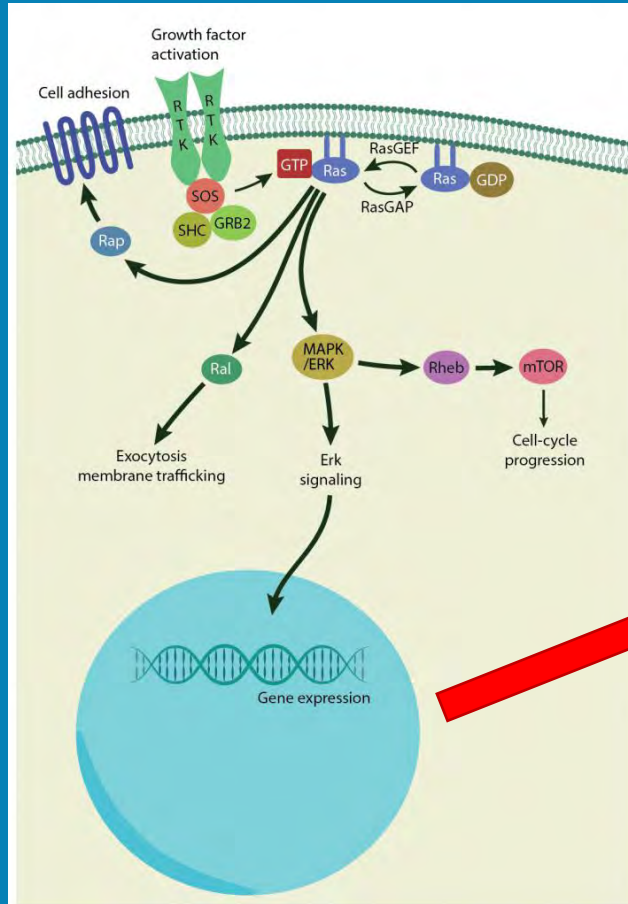
Growth – not uncommon but worth discussing



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Uncommon things in NF1



How does NF1 cause shorter bones



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Uncommon things in NF1

PRECOCIOUS PUBERTY



DEVELOPMENT OF
SECONDARY SEXUAL
CHARACTERISTICS
BEFORE:

- AGE 8 IN GIRLS
- AGE 9 IN BOYS



MEDSKL



E 13

Puberty



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Uncommon things in NF1

Vascular System

Causes:

- Idiopathic / Essential
 - Most cases
- Renal artery Stenosis
 - Does not respond to medication
 - Needs the blood supply to the kidney to be fixed
 - More common in children with high blood pressure
- Pheochromocytoma
 - Secretes the “fight/flight” hormone
 - Not cancer but does need to be treated

Hypertension = High Blood Pressure



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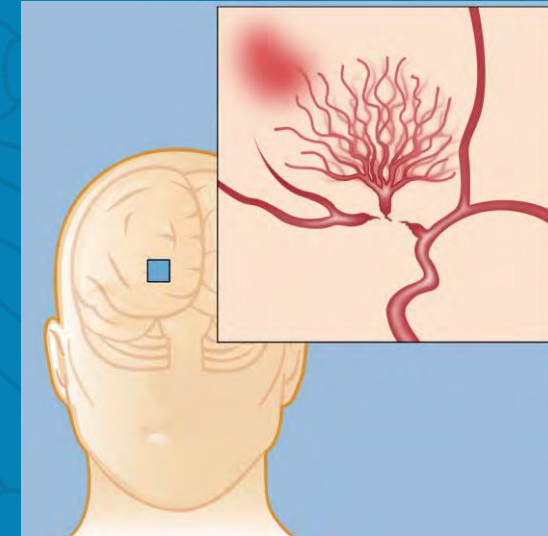
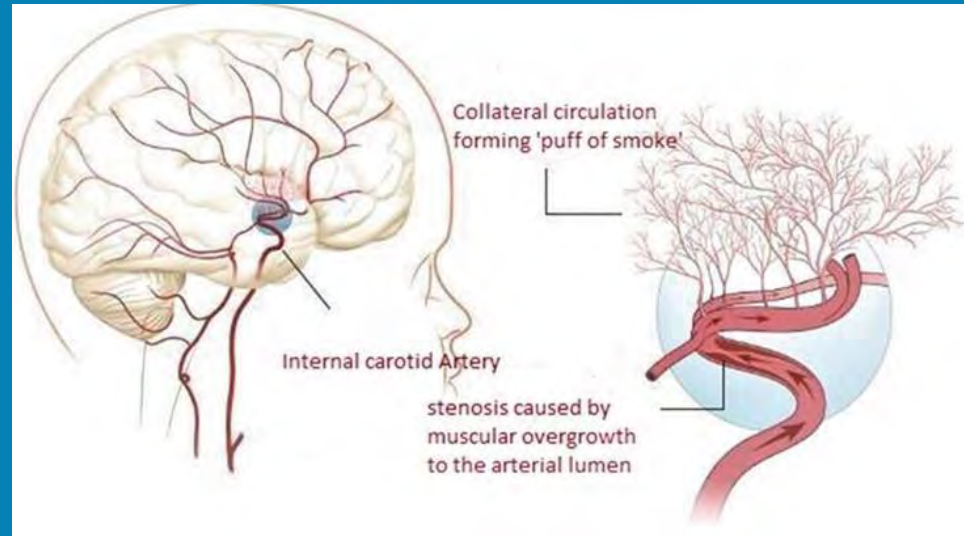
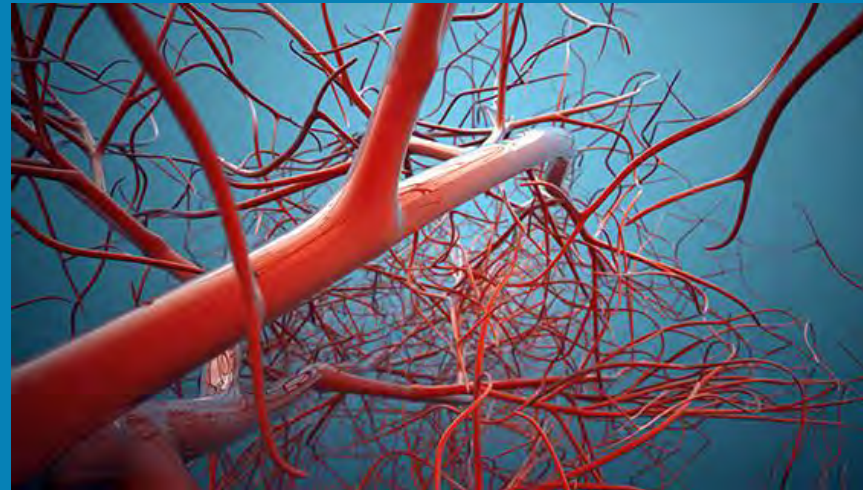


Uncommon things in NF1

Can be found anywhere – most are not important

Vascular System

- Brain:
 - Cerebrovascular malformations
 - Moyamoya ~ “puff of smoke” - increased risk for stroke



Abnormal blood vessels



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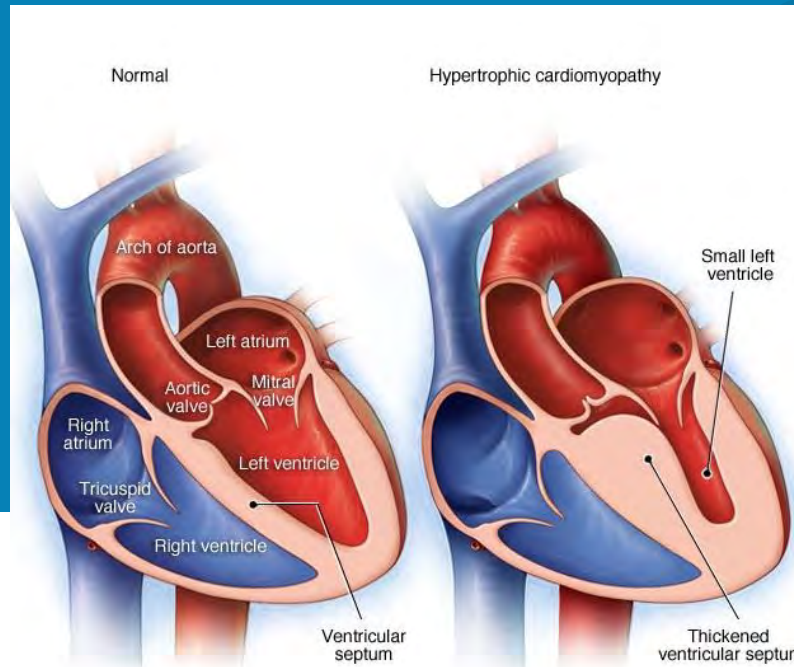


Uncommon things in NF1

Heart



- Valves:
 - Pulmonic stenosis
- Function:
 - Cardiomyopathy (heart muscles enlarge)



More common with
NF1 gene deletions

Uncommon things in NF1

Tumors



- Leukemia
 - Rare but more common in NF1 than general population
- other tumors
 - Rhabdomyosarcomas
 - Pheochromocytomas
 - Gastrointestinal stromal tumors
 - Glomus tumors
 - Retinal vasoproliferative tumors
 - Women with NF1 are at higher risk for developing breast cancer before age 50

Tumors



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Transition Planning



Pediatric Care

- child/family centered
- parental involvement /ensure compliance
- multidisciplinary

Adult Care

- patient centered
- autonomy / independence
- physician directed (referrals to other specialists)

Know your resources and ask for help



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Transition to Adult Care

- Know your diagnosis
 - Gives confidence
 - Help direct appropriate care
 - Rare but more common in NF1 than general population
- Manage Health Care needs
 - Know the symptoms
 - Know the medications
 - Know who to call if you have problems
- Learn about your health insurance
 - Understand benefits and social security
- Educate about additional pressures of life outside of your family home

Why is it important to think about



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Resources

Got Transition : www.gottransition.org

Family Voices of Wisconsin : www.familyvoicesofwisconsin.com

WI Dept of Health : www.dhs.Wisconsin.gov/cyshcn/regionalcenters

NF Collective: www.nfcollective.org/resources

CW symposium :



<https://www.youtube.com/watch?v=ekL8y85zjyw>

The Guide for NF Patients and Caregivers Transitioning (PACT) to Adult Care



The Guide for NF Patients and Caregivers Transitioning (PACT) to Adult Care is a resource to help patients and caregivers with the process of transitioning from pediatric to adult health care. Based upon best practice research and longitudinal experience from members of the NF Collective, this guide offers specific suggestions for precise periods of development during NF patient's maturation. Stressing increased patient autonomy while still honoring the vital partnerships with caregivers, this guide aims to be an effective resource for the NF community to best promote a smooth transition into adulthood for all impacted. In addition to the detailed suggestion sections, this guide also provides individuals with online resources and a glossary of terms to present a most comprehensive tool that, we hope, families will find of great value.

DOWNLOAD

NF Collective Webinar: Transitioning to Adult NF Care



On February 24, 2021 the NF Collective hosted an inspirational webinar of young adult NF patients who shared personal experiences and advice on transitioning to adult NF care. This event was held in conjunction with the release of "The Guide for NF Patients and Caregivers Transitioning (PACT) to Adult Care", a new resource for the NF community put together by the NF Collective. In this webinar, you'll hear personal testimonials, be inspired to better navigate the stressful but exhilarating journey into NF young adulthood, and reflect on the sharing of personal challenges and triumphs of our esteemed panel of NF young adults.

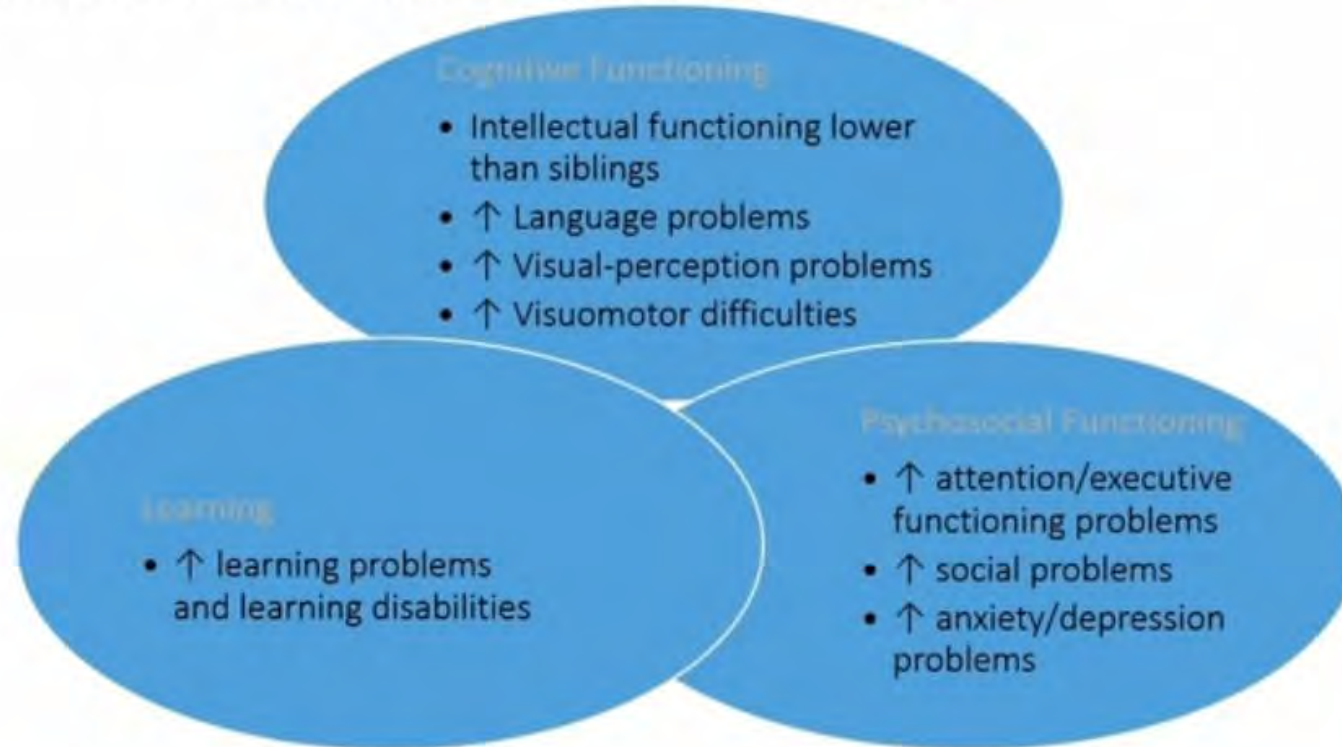
WATCH WEBINAR



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Common Cognitive and Psychosocial Difficulties in NF1



(e.g., Cutting et al., 2000, 2004, Dilts et al., 1996, Schrimsher et al., 2003)

- High level of variability
- Not a consistent pattern



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