2019 ACOI BOARD REVIEW

GENETICS

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Founder
Roundocity
Hospitalist

Chromosomal Abnormalities

nefelter Syndrome, Turner Syndrome, Fragile X-linked mental re

Chromosomal

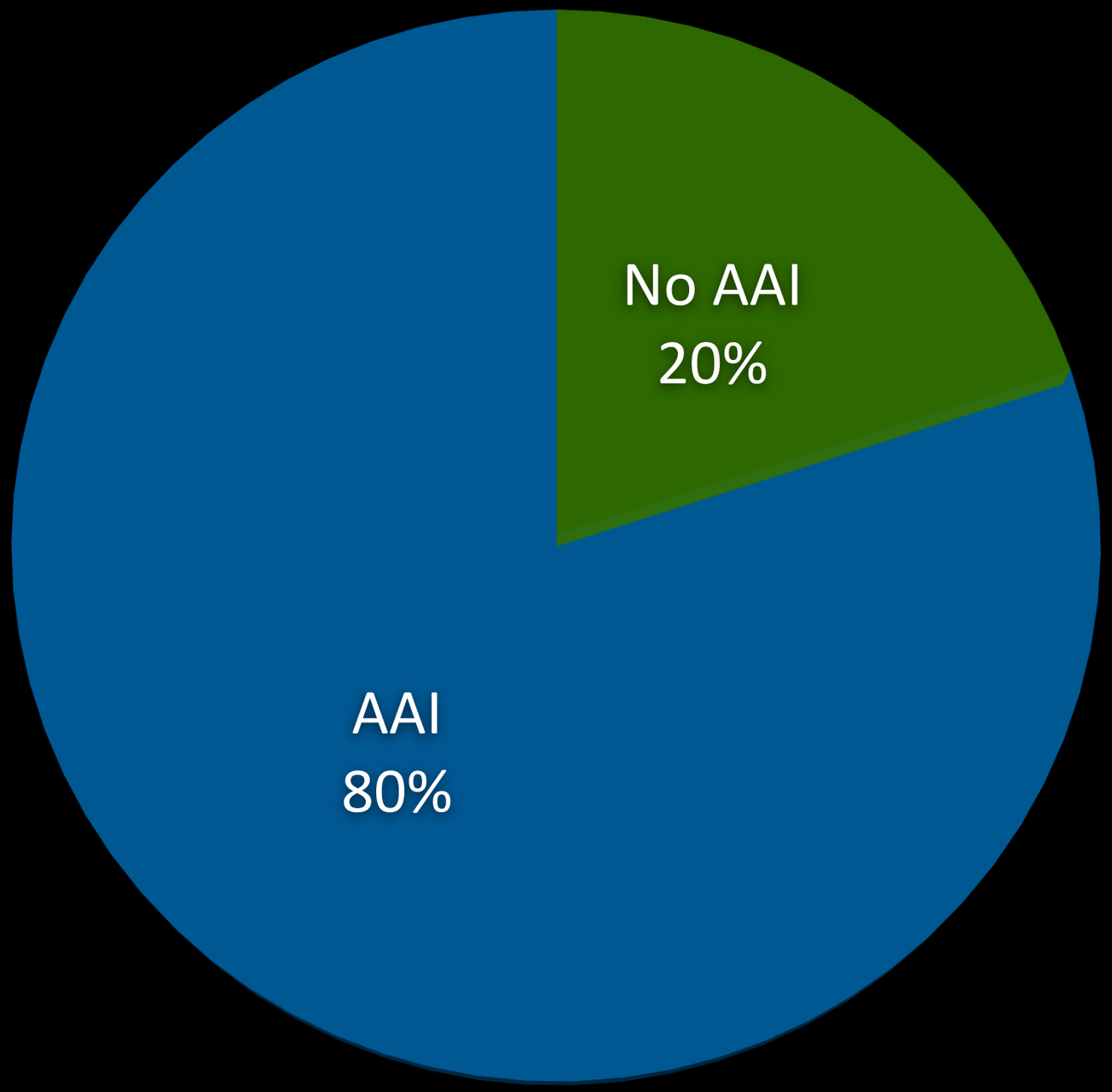
Down Syndrome (trisomy 21)

Increased ALL

Early Alzheimer Dementia

VSD & AV Canal Defects

ATLANTOAXIAL INSTABILITY (AAI) IN DOWN SYNDROME



Int Orthop. Aug 2006; 30(4): 284–289.
Published online Mar 7, 2006. doi: 10.1007/s00264-005-0070-y

cervical spine radiographs should be ordered to assess for odontoid-axial stability in patients with rheumatoid arthritis & Down Syndrome

Chromosomal

Klinefelter (47,XXY)

Increased Risk for Germ Cell Tumors
Infertility/Small Testes
Tall/Eunuchoid Habitus

Chromosomal

Turner (45, XO)

Normal Mentation

Associated with Coarction/Bicuspid Aorta

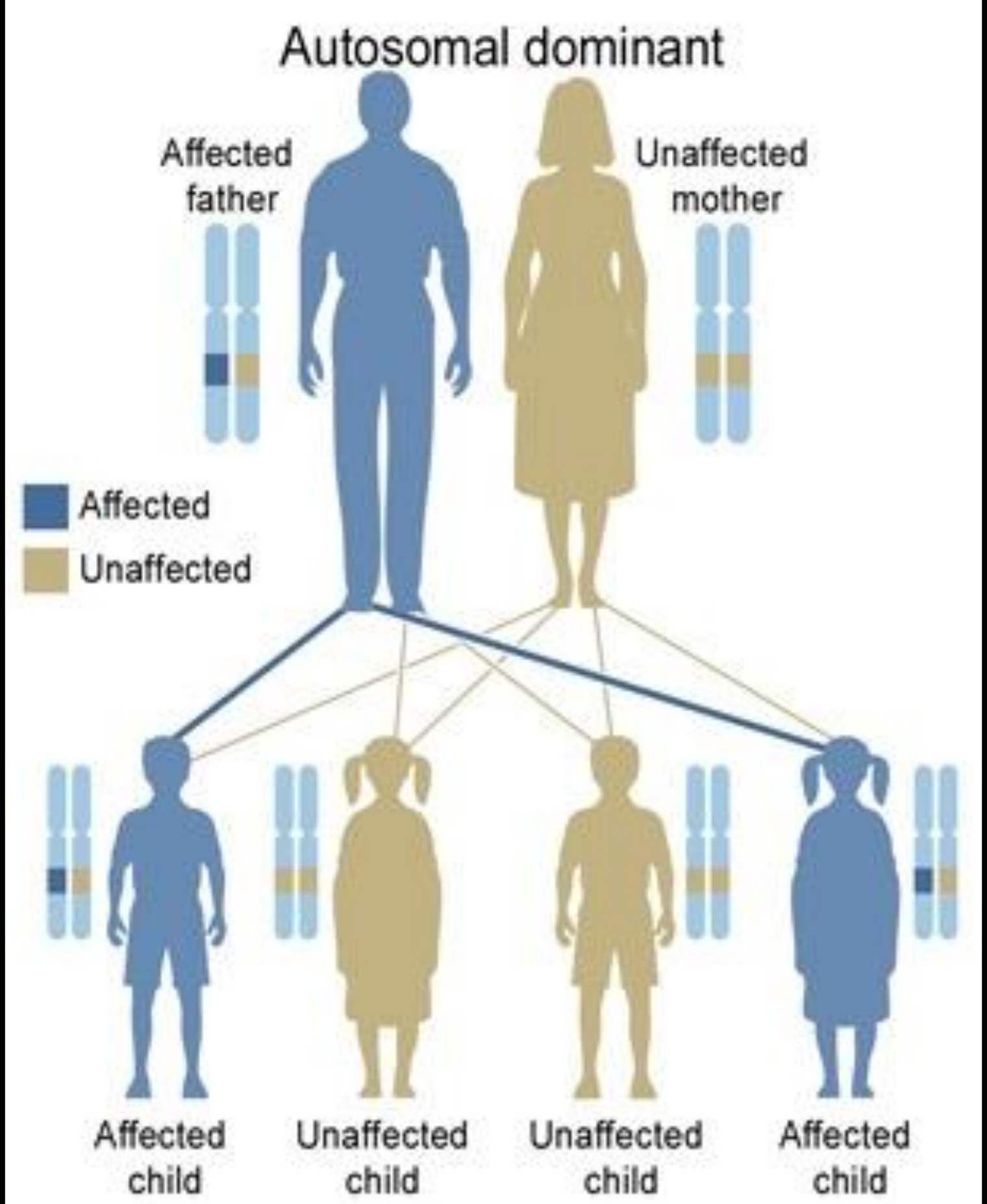
Chromosomal

Fragile X-linked mental retardation syndrome

Look normal or with
thin face/large jaw & ears/enlarged testes

Autosomal Dominant

hypercholesterolemia), Lynch syndrome, multiple endocrine ne



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Autosomal Dominant

Marfan Syndrome

MVP & Ascending Aorta Dilatation

Ectopic Lens

Fibrillin Gene

Autosomal Dominant

Ehlers-Danlos Syndrome

Collagen Defect - Hypermobility/Fragile Skin

MVP/Arterial Aneurysms/Organ Ruptures

Autosomal Dominant

BRCA mutations

50-80% will develop breast ca

BRCA 2 associated with male breast ca

Autosomal Dominant

Hereditary Spherocytosis

Cytoskelatin Defect

Pigmented Gallstones/Hemolysis

Splenectomy can be helpful

Dominant

Huntington Disease

Autosomal

HTT Gene Mutation/CAG nucleotide repeat

Autosomal Dominant

Hereditary Angioedema

No Urticaria

PEARL: PERFORM GENETIC COUNSELING (1HOUR) BEFORE TESTING (E.G. HUNTINGTON FAMILY HX)

Autosomal Dominant

Lynch syndrome

Hereditary Non-polyposis Colon CA

5% of Colon CA

Autosomal Dominant

Multiple Endocrine Neoplasias types I, IIA, and IIB

TYPE	ASSOCIATION	COMMENTS
1	TUMORS OF PARATHYROID, PITUITARY, PANCREAS	HYPERPARATHYROIDISM IS MOST COMMON SIGN
2 (A, B, FAMILIAL MEDULLARY THYROID CARCINOMA)	MEDULLARY THYROID CARCINOMA	
4	SIMILAR TO TYPE 1	

Autosomal Dominant

Polycystic Kidney Disease

PKD 1 or 2 Gene Mutations

Strong Association with Intracranial Aneurysms

von Willebrand Disease

Autosomal Dominant

Women are more commonly diagnosed (menses)

Autosomal Dominant

Hypertrophic Cardiomyopathy

Murmur Decreases with Hand Grip/
Louder with Valsalva

Autosomal Dominant

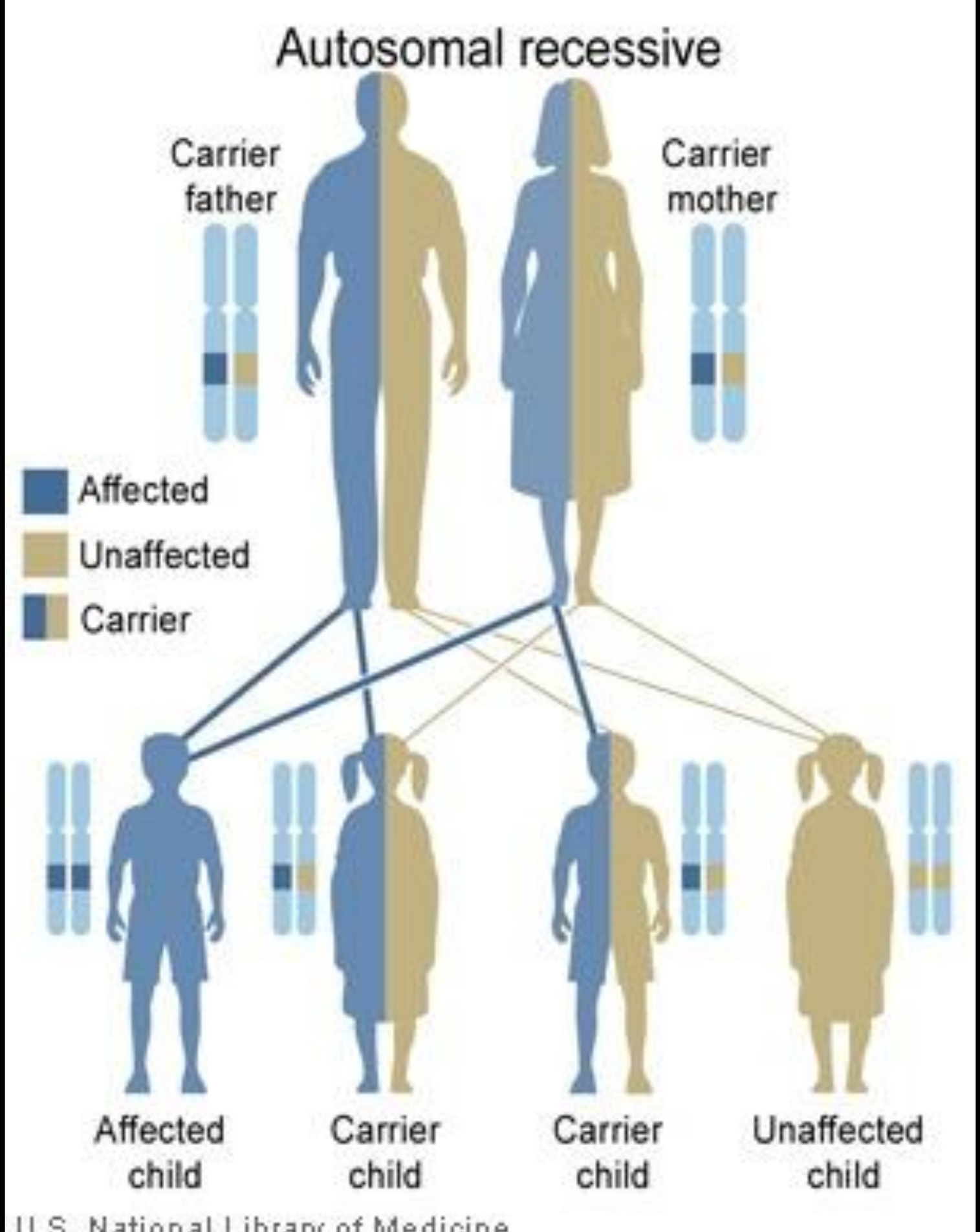
Myotonic Dystrophy

Triplet repeat (myotonin protein kinase gene)
Prolonged rhythmic discharges on EEG

A type of muscular dystrophy
Onset in 2nd/3rd decade

Autosomal Recessive

cystic fibrosis, hemochromatosis, sickle cell anemia, the thalass


Autosomal recessive
Carrier father
Carrier mother
Affected
Unaffected
Carrier
Affected child
Carrier child
Carrier child
Unaffected child
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Autosomal Recessive

α 1 -Antitrypsin Deficiency

Basilar Emphysema

Autosomal Recessive

Cystic Fibrosis

Think Intussusception with Abdominal Pain

Burkholderia cepacia in addition to pseudomonas

Autosomal Recessive

Hemochromatosis

"Hook-Like Osteophytes" in 2nd & 3rd MCP Joints

HFE gene testing if transferrin saturation>45%

Autosomal Recessive

Sickle Cell Anemia

VAL substituted for GLU

Recessive

Autosomal

Thalassemia

Alpha or Beta - Named after Missing Part

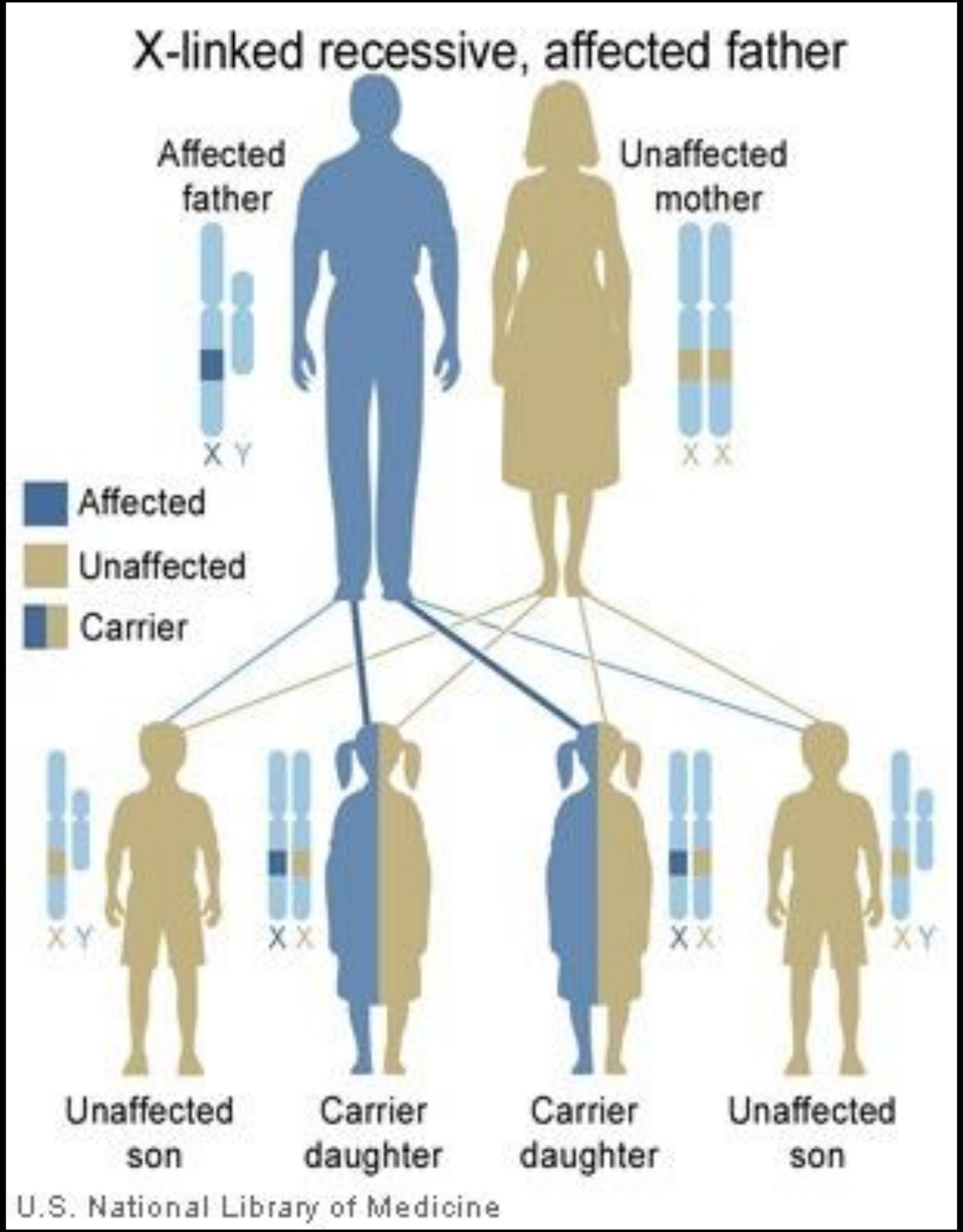
Autosomal Recessive

Wilson Disease

Mutations of the ATP7B gene

X-Linked Recessive

G6PD, Hemophilia A & B


X-linked recessive, affected father
Affected father
Unaffected mother
X Y
X X
Affected
Unaffected
Carrier
X Y
X X
X X
X Y
Unaffected son
Carrier daughter
Carrier daughter
Unaffected son
U.S. National Library of Medicine

Recessive

X-Linked

G6PD

Shorter RBC Lifespan

Occurs in 10% of African American Males

Avoid: NSAID, ASA, Nitrofurantoin,
Quinine, Sulfa, Antimalarials

Recessive

X-Linked

Hemophilia A & B

Almost exclusively in males

Typically diagnosed at a Very Young Age

Mitochondrial Mutations

ny, episodes of lactic acidosis, and stroke (MELAS), myoclonic ep

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