2019 ACOI BOARD REVIEW

GENETICS

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Idaho College of Osteopathic Medicine

ICOM
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Chromosomal Abnormalities

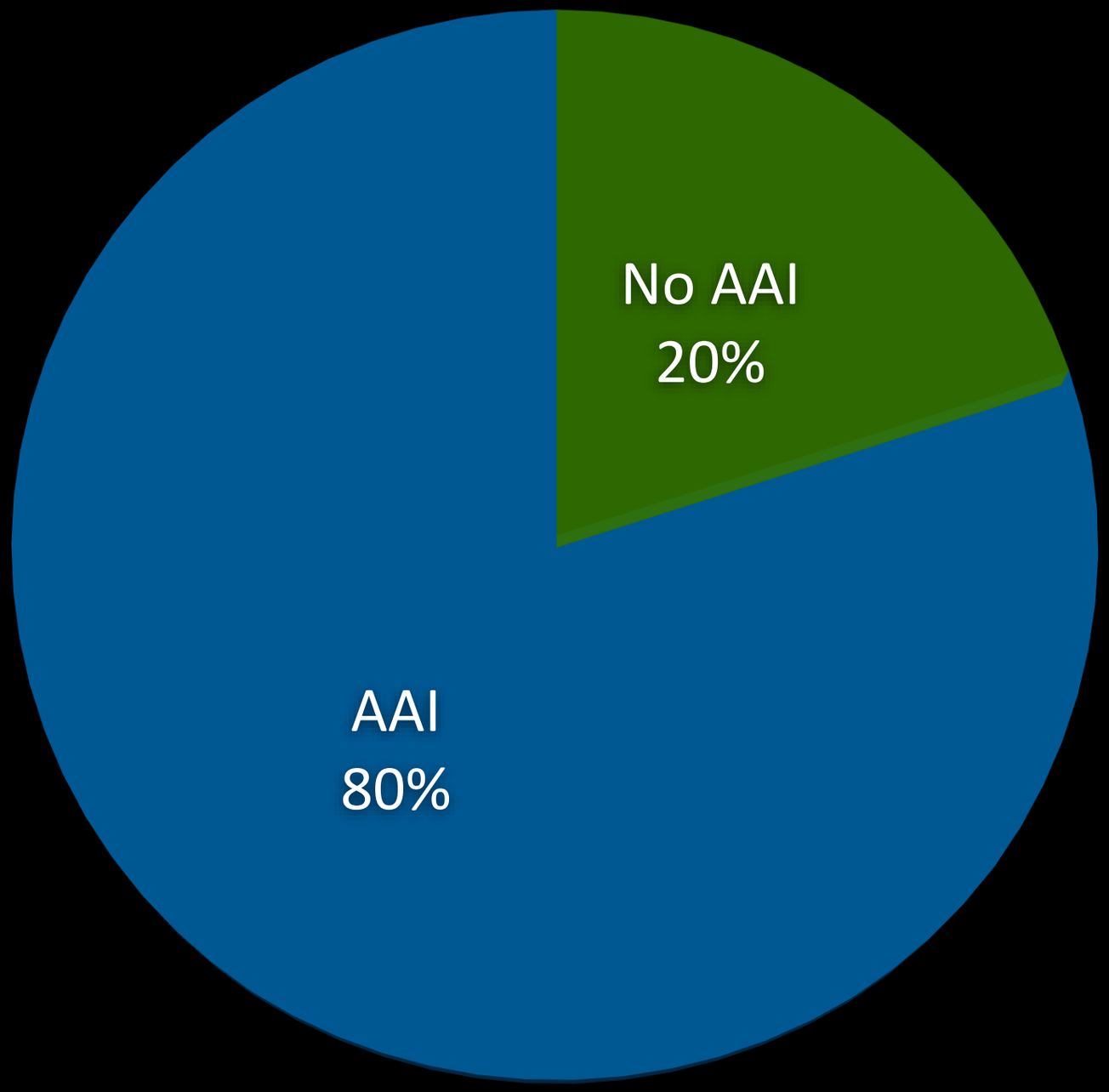
nefelter Syndrome, Turner Syndrome, Fragile X-linked mental re

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

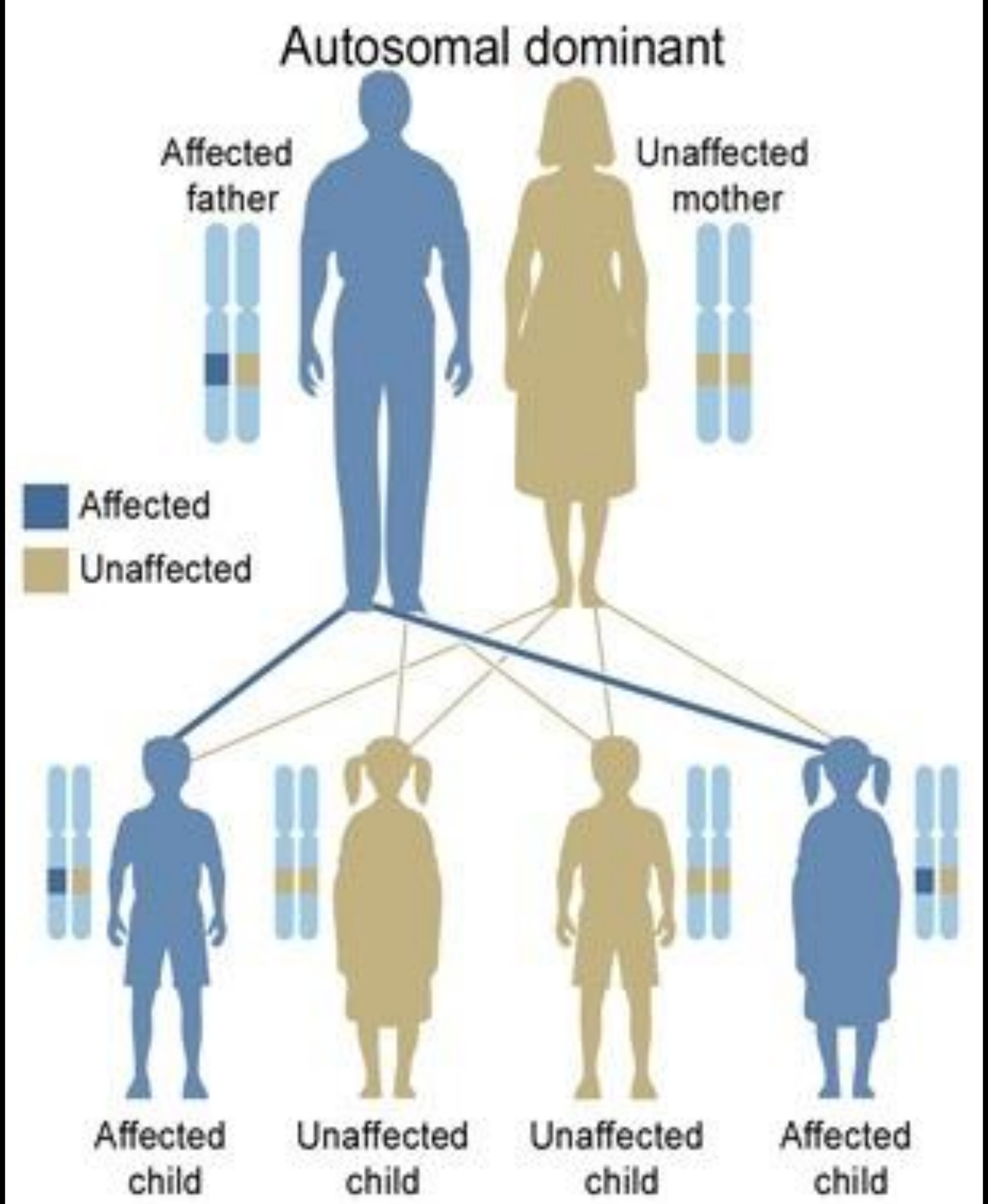
Chromosomal

Fragile X-linked mental retardation syndrome

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

Autosomal Dominant

ency (familial hypercholesterolemia), Lynch syndrome, multiple



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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

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

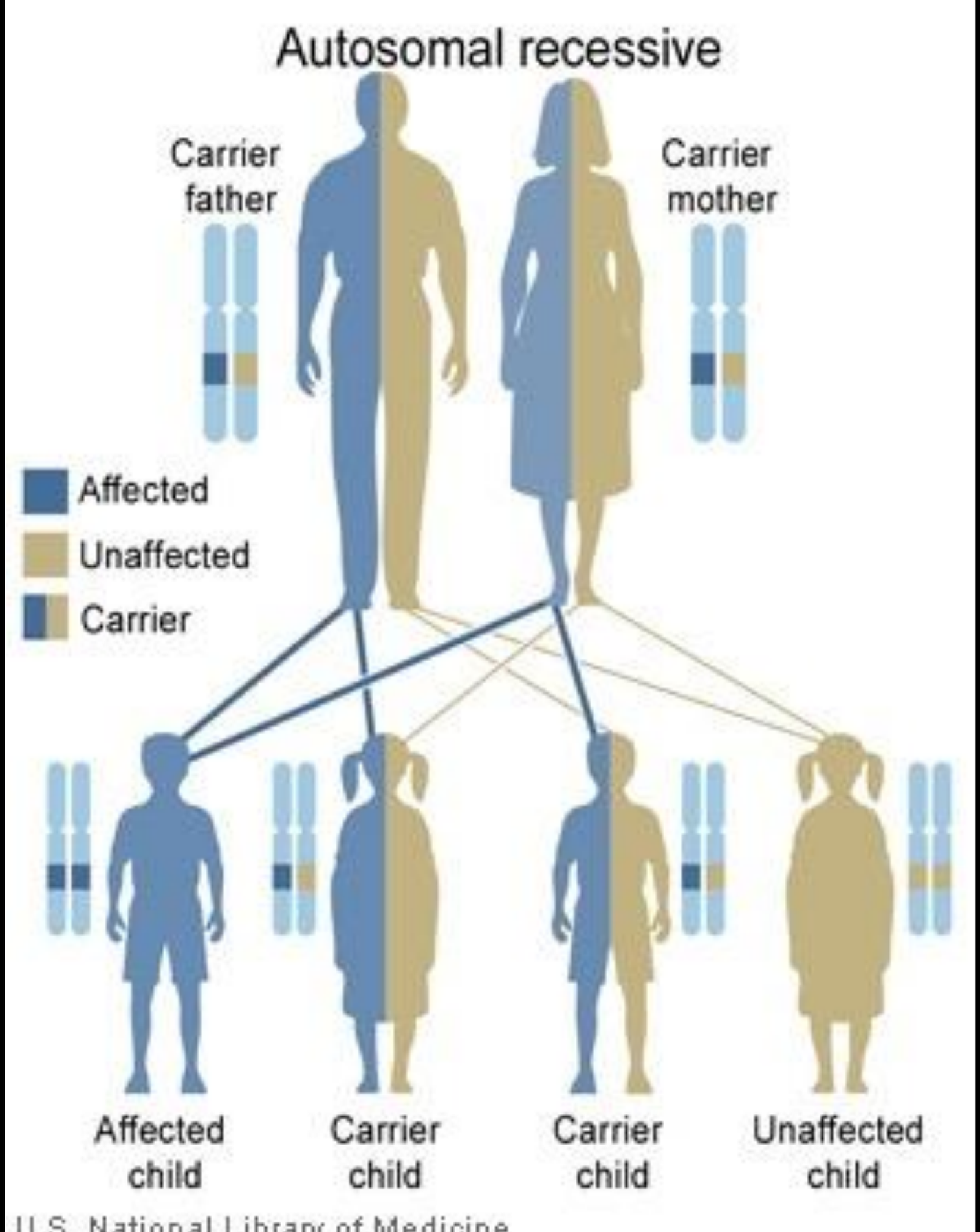
Autosomal Dominant

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

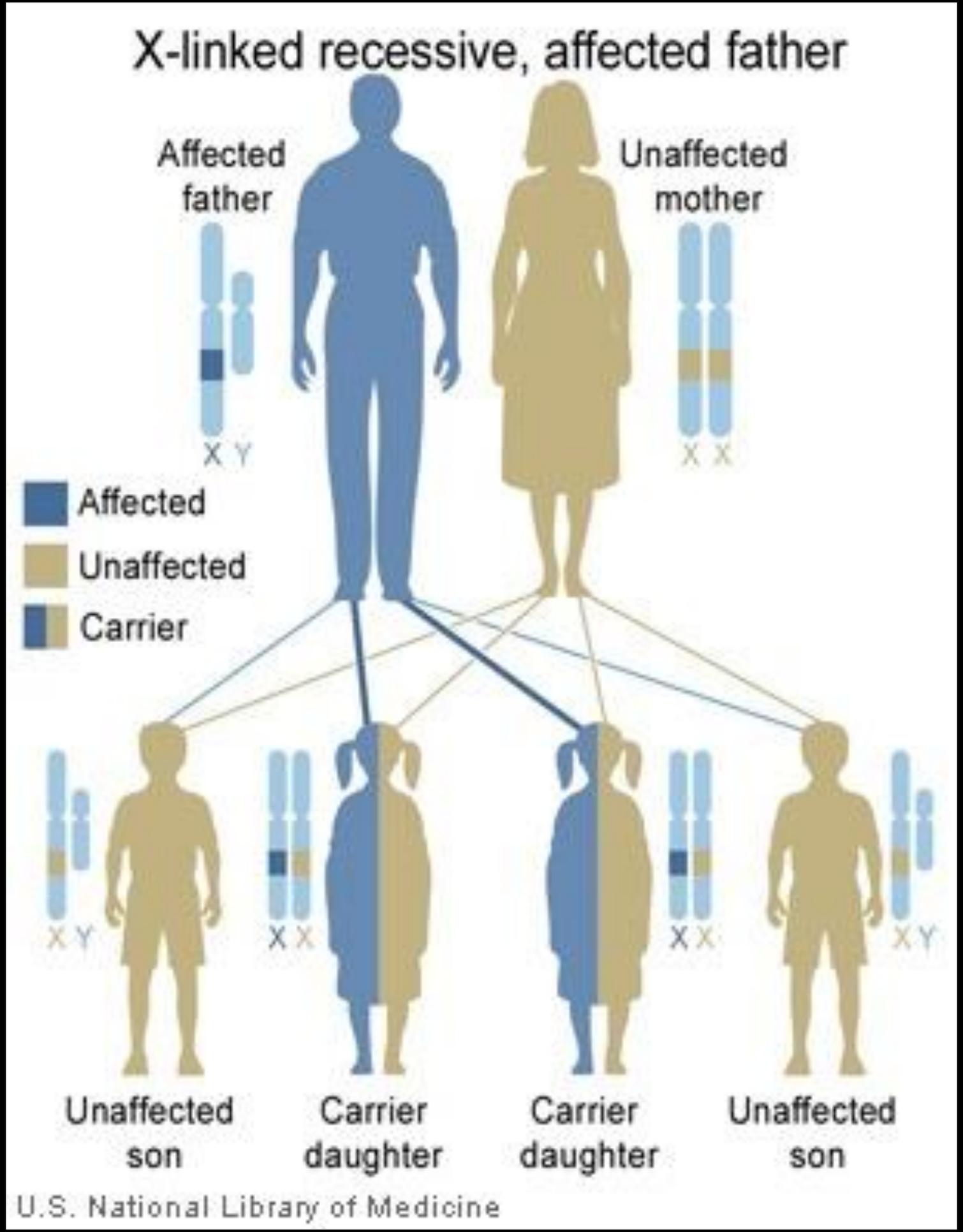
Autosomal Recessive

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
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X-Linked Recessive

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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