2017 ACOI BOARD REVIEW

GENETICS

Robert Hasty, DO, FACOI, FACP

Founding Dean & Chief Academic Officer
Idaho College of Osteopathic Medicine - Applicant Status

Proposed
IDAHO
College of Osteopathic Medicine

Chromosomal Abnormalities

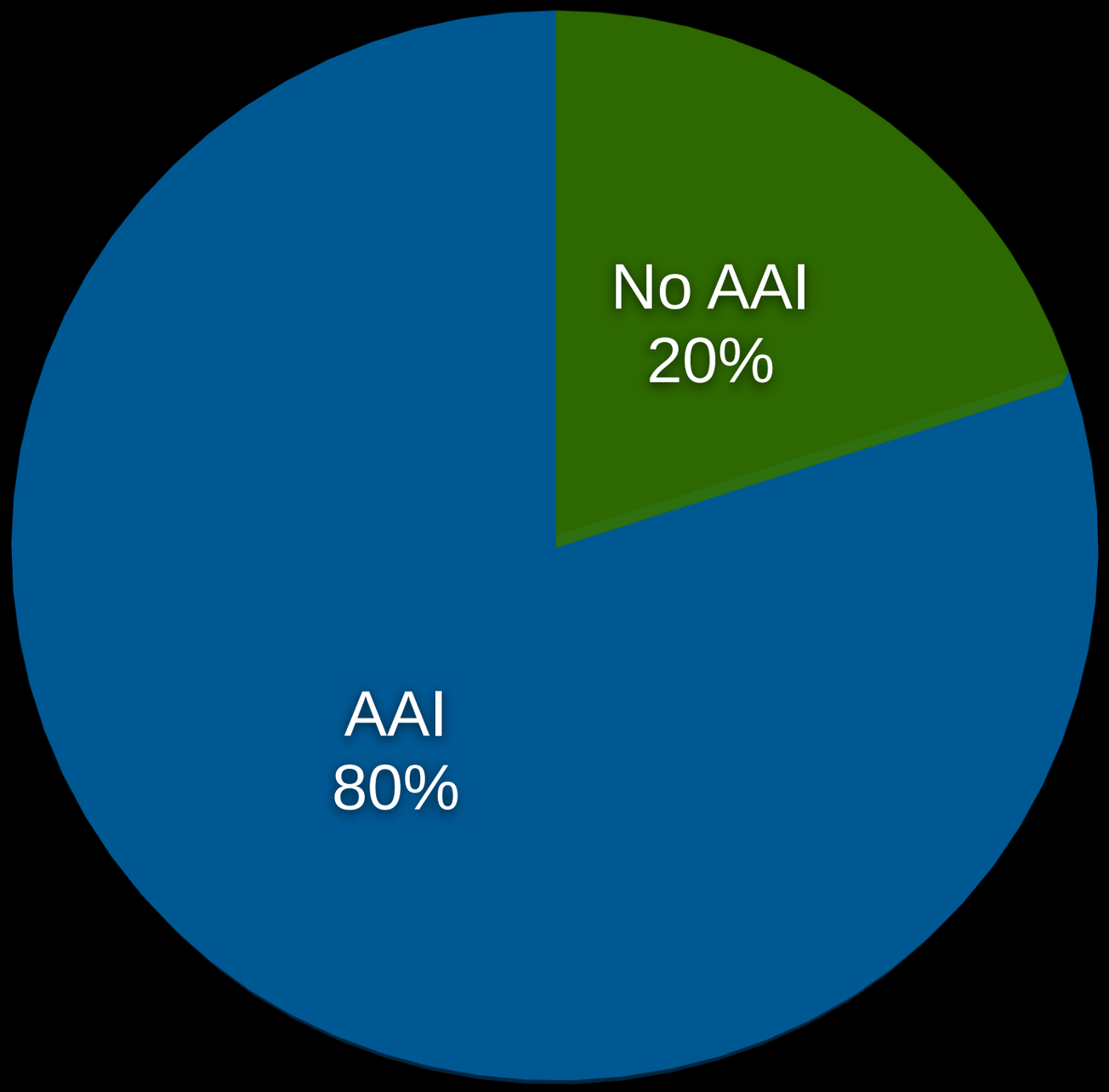
felter Syndrome, Turner Syndrome, Fragile X-linked menta

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

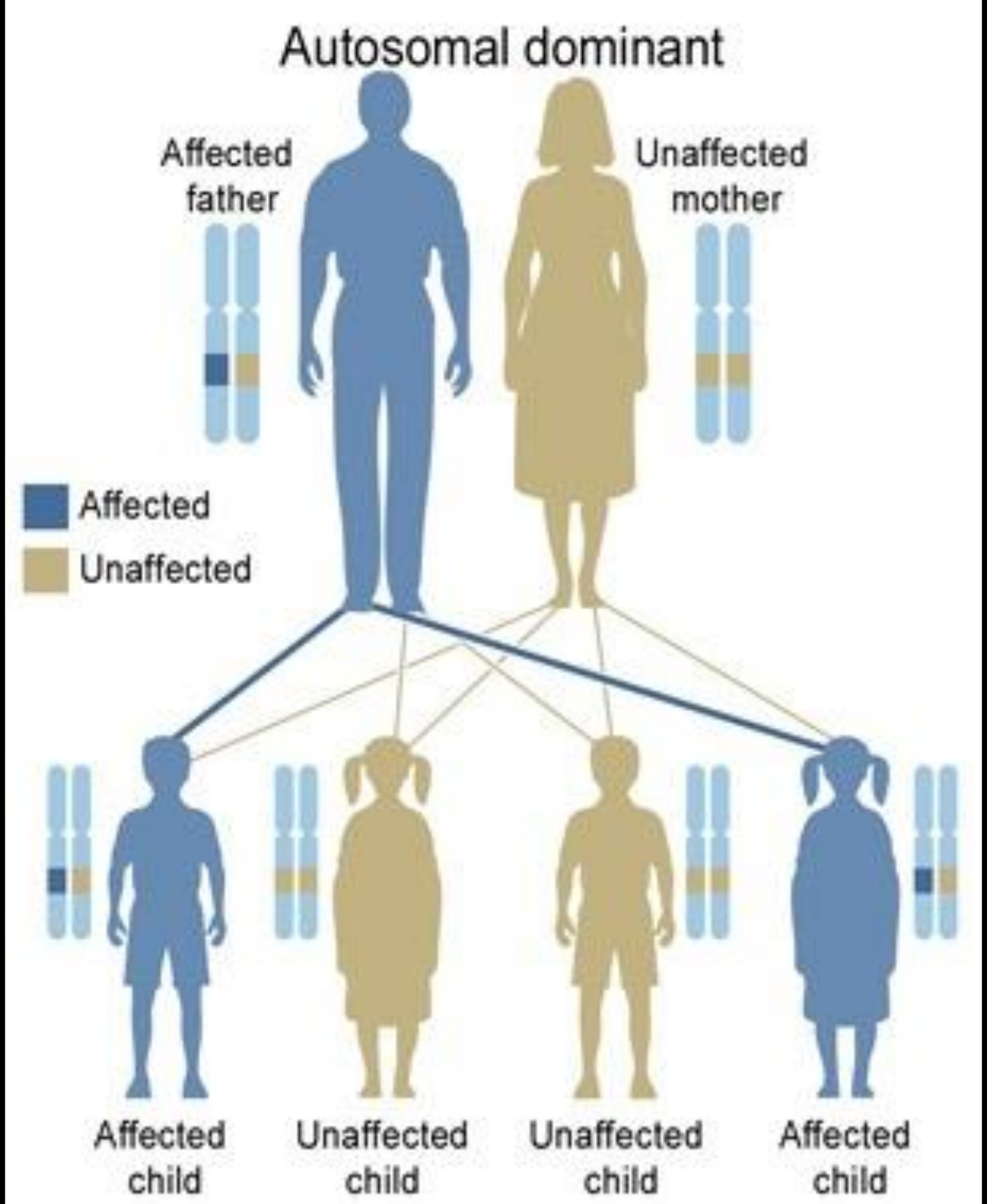
Chromosomal

Fragile X-linked mental retardation syndrome

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

Autosomal Dominant

y (familial hypercholesterolemia), Lynch syndrome, multipl



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Marfan Syndrome

Autosomal Dominant

MVP & Ascending Aorta Dilatation

Ectopic Lens

Fibrillin Gene

Ehlers-Danlos Syndrome

Autosomal Dominant

Collagen Defect - Hypermobility/Fragile Skin
MVP/Arterial Aneurysms/Organ Ruptures

BRCA mutations

Autosomal Dominant

50-80% will develop breast ca

BRCA 2 associated with male breast ca

Hereditary Spherocytosis

Autosomal Dominant

Cytoskelatin Defect

Pigmented Gallstones/Hemolysis

Splenectomy can be helpful

Huntington Disease

Autosomal
Dominant

HTT Gene Mutation/CAG nucleotide repeat

Lynch syndrome

Autosomal Dominant

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	

Polycystic Kidney Disease

Autosomal Dominant

PKD 1 or 2 Gene Mutations

Strong Association with Intracranial Aneurysms

von Willebrand Disease

Autosomal
Dominant

- Women are more commonly diagnosed (menses)

Hypertrophic Cardiomyopathy

Autosomal Dominant

Murmur Decreases with Hand Grip

Myotonic Dystrophy

Autosomal Dominant

Triplet repeat (myotonin protein kinase gene)

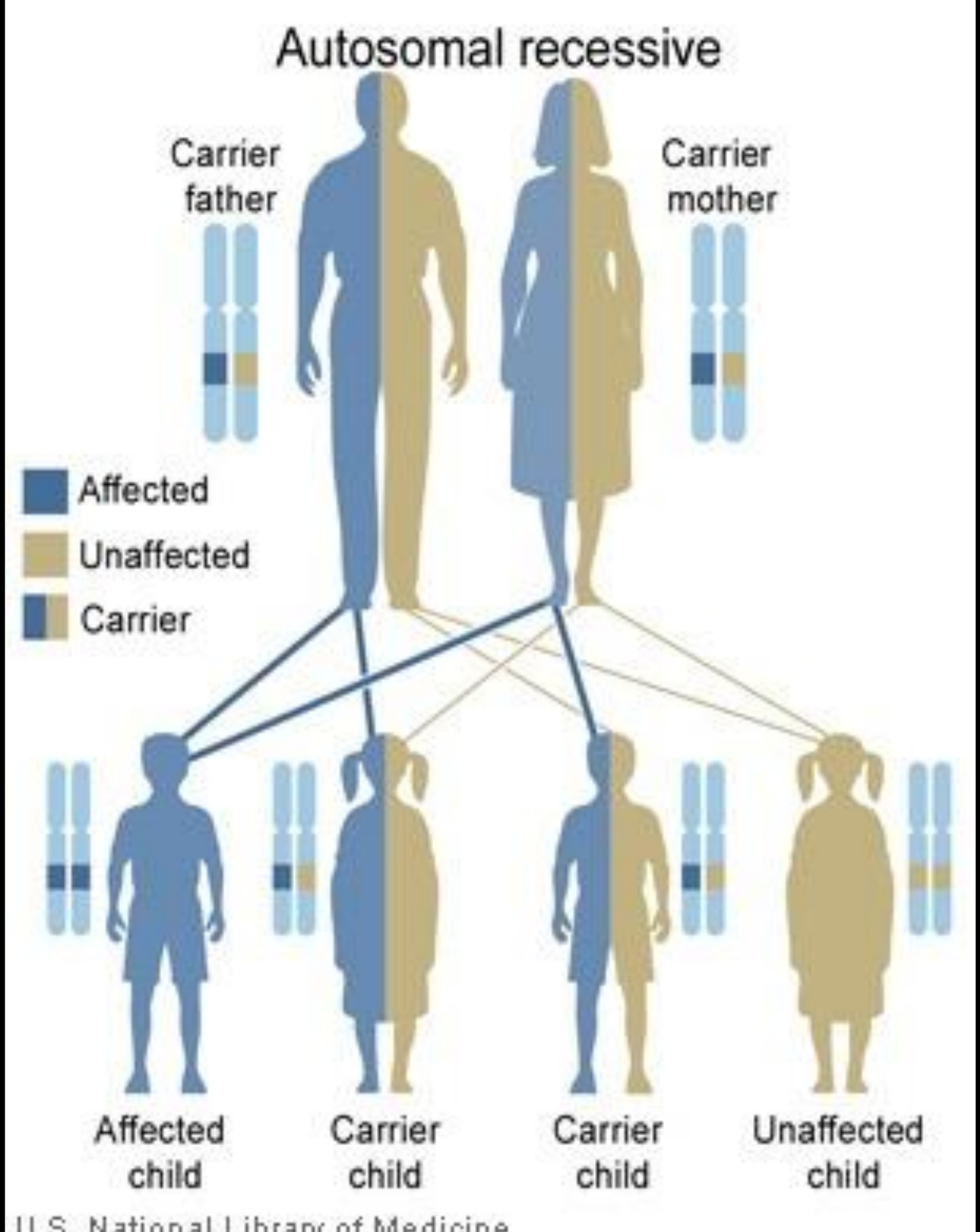
Prolonged rhythmic discharges on EEG

A type of muscular dystrophy

Onset in 2nd/3rd decade

Autosomal Recessive

stic fibrosis, hemochromatosis, sickle cell anemia, the thala


Autosomal recessive
Carrier father
Carrier mother
Affected
Unaffected
Carrier
Affected child
Carrier child
Carrier child
Unaffected child
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α 1 -Antitrypsin Deficiency

Autosomal
Recessive

Basilar Emphysema

Cystic Fibrosis

Autosomal Recessive

Think Intussusception with Abdominal Pain

Burkholderia cepacia in addition to pseudomonas

Hemochromatosis

Autosomal Recessive

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

HFE gene testing if transferrin saturation>45%

Sickle Cell Anemia

Autosomal
Recessive

VAL substituted for GLU

Thalassemia

Autosomal Recessive

Alpha or Beta - Named after Missing Part

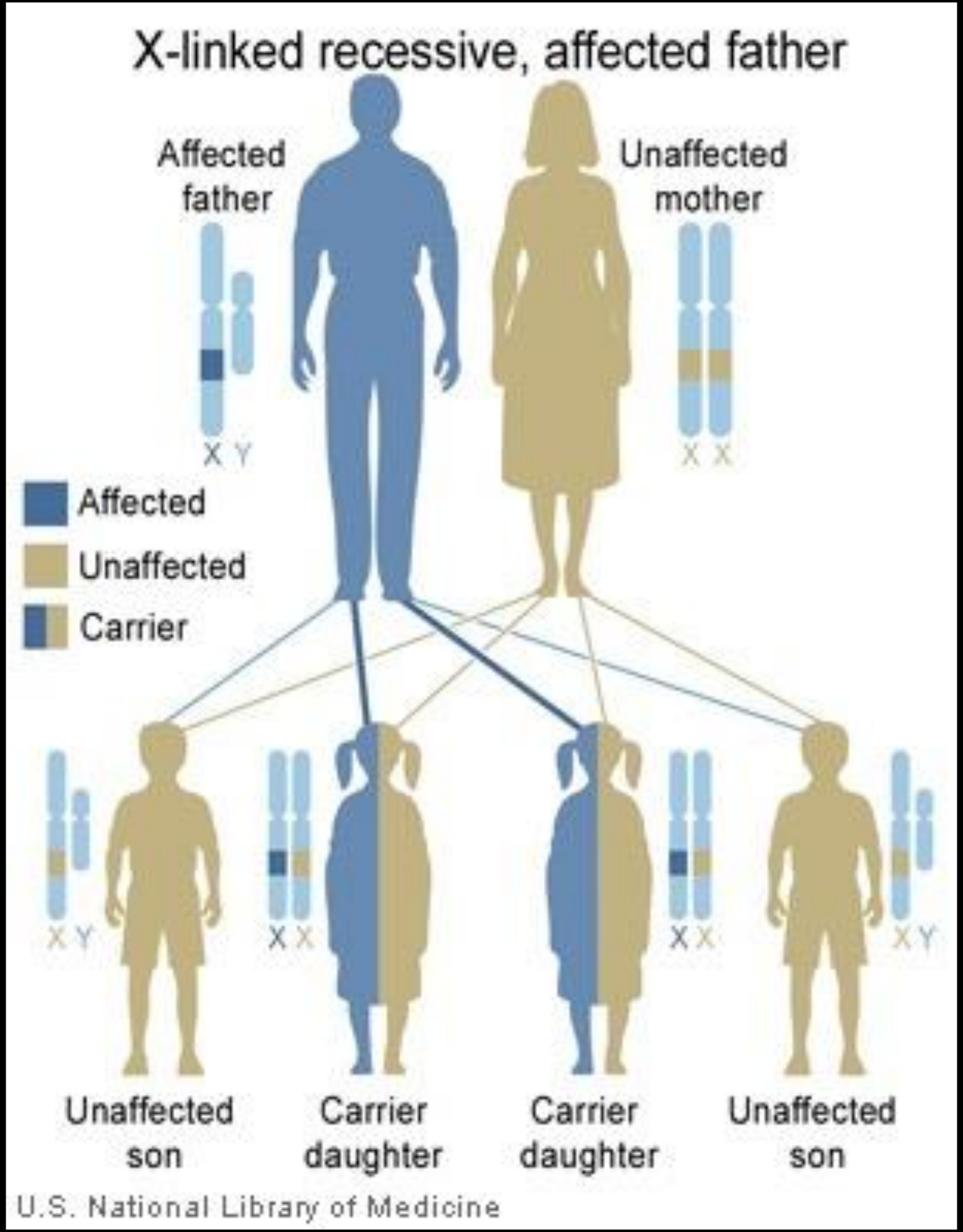
Wilson Disease

Autosomal Recessive

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

X-Linked Recessive

Hemophilia A & B

Almost exclusively in males

Typically diagnosed at a Very Young Age

Mitochondrial Mutations

isodes of lactic acidosis, and stroke (MELAS), myoclonic

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