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2016 ACOI BOARD REVIEW

GENETICS
Chromosomal Abnormalities

Klinefelter Syndrome, Turner Syndrome, Fragile X-linked mental retardation syndrome
Down Syndrome (trisomy 21)

- Increased ALL
- Early Alzheimer Dementia
- VSD & AV Canal Defects
ATLANTOAXIAL INSTABILITY (AAI) IN DOWN SYNDROME

- AAI: 80%
- No AAI: 20%

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cervical spine radiographs should be ordered to assess for odontoid-axial stability in patients with rheumatoid arthritis & Down Syndrome
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
Fragile X-linked mental retardation syndrome

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

- Familial hypercholesterolemia
- Lynch syndrome
- Multiple endocrine neoplasias types I, IIA, and IIB

(familial hypercholesterolemia), Lynch syndrome, multiple endocrine neoplasias types I, IIA, and IIB
Autosomal Dominant

Marfan Syndrome

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

50-80% will develop breast cancer

BRCA 2 associated with male breast cancer
Hereditary Spherocytosis

Autosomal Dominant

Cytoskelatin Defect
Pigmented Gallstones/Hemolysis
Splenectomy can be helpful
Huntington Disease

Autosomal Dominant

HTT Gene Mutation/CAG nucleotide repeat
Autosomal Dominant

Lynch syndrome

Hereditary Non-polyposis Colon CA
5% of Colon CA
# Multiple Endocrine Neoplasias types I, IIA, and IIB

<table>
<thead>
<tr>
<th>TYPE</th>
<th>ASSOCIATION</th>
<th>COMMENTS</th>
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<tbody>
<tr>
<td>1</td>
<td>TUMORS OF PARATHYROID, PITUITARY, PANCREAS</td>
<td>HYPERPARATHYROIDISM IS MOST COMMON SIGN</td>
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<td>2 (A, B, FAMILIAL MEDULLARY THYROID CARCINOMA)</td>
<td>MEDULLARY THYROID CARCINOMA</td>
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<td>4</td>
<td>SIMILAR TO TYPE 1</td>
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Polycystic Kidney Disease

Autosomal Dominant

PKD 1 or 2 Gene Mutations
Strong Association with Intracranial Aneurysms
von Willebrand Disease

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
Antitrypsin deficiency, cystic fibrosis, hemochromatosis, sickle cell anemia, the thalas...
α 1 -Antitrypsin Deficiency

Autosomal Recessive

Basilar Emphysema
Cystic Fibrosis

Think Intussusception with Abdominal Pain

*Burkholderia cepacia* in addition to pseudomonas
Hemochromatosis

"Hook-Like Osteophytes" in 2nd & 3rd MCP Joints
HFE gene testing if transferrin saturation > 45%
Sickle Cell Anemia

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
G6PD

Shorter RBC Lifespan
Occurs in 10% of African American Males
Avoid: NSAID, ASA, Nitrofurantoin, Quinine, Sulfa, Antimalarials
Hemophilia A & B

Almost exclusively in males
Typically diagnosed at a Very Young Age
Mitochondrial Mutations

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