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Founding Dean & Chief Academic Officer
Idaho College of Osteopathic Medicine
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%

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 Coarctation/Bicuspid Aorta
Fragile X-linked mental retardation syndrome

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

- Receptor deficiency (familial hypercholesterolemia), Lynch syndrome, multiple endocrine neoplasias types I, IIA, and IIB, poly...
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
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
Huntington Disease

- Autosomal Dominant
- HTT Gene Mutation/CAG nucleotide repeat
PEARL: PERFORM GENETIC COUNSELING (1 HOUR) BEFORE TESTING (E.G. HUNTINGTON FAMILY HX)
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>HYPERPARATHYROIDIS M 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

Autosomal Dominant

Women are more commonly diagnosed (menses)
Autosomal Dominant

Hypertrophic Cardiomyopathy

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 thalassemias, and Wilson disease are examples of autosomal recessive traits.
Autosomal recessive

- Carrier father
- Carrier mother

- Affected
- Unaffected
- Carrier

- Affected child
- Carrier child
- Carrier child
- Unaffected child
α 1 -Antitrypsin Deficiency

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
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 e
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Idaho College of Osteopathic Medicine - Applicant Status