Pediatric manifestations of Adult conditions

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Disclosures

I have no disclosures or financial investments in any of the conditions or medications I will be discussing.

Objectives

- Case presentation and discussion of autoimmune illness in peds
  - Crohn's disease, JIA, Autoimmune thyroiditis, Lupus

Case presentation and discussion of inherited metabolic disorder

Case presentation and discussion of inherited illness affecting lung and liver

Case presentation and discussion of inherited pulmonary condition

Case presentation and discussion of inherited neurocutaneous disorder

Case presentation and discussion of inherited bleeding disorder

Case presentation and discussion of connective tissue disorder

Case presentation and discussion of abnormal growth

Case presentation and discussion of abnormal bone mineralization

Case presentation and discussion of chronic headache with inherited condition

Case presentation and discussion of inherited abnormality in glucose metabolism

Case 1

Case presentation AS 14 year old

Presented on 03/11/2024 with severe abdominal pain, persistent vomiting, no blood or bile and watery diarrhea, with 30 lb weight loss over the last 4 months. She present with acute pain to Er and a ct scan was done showing. She had been having morning joint pain. She had developed a facial rash.

Bowel wall thickening of the mid and distal ileum along with the
distal sigmoid colon with associated moderate ascites most consistent
with infectious enteritis/colitis.

Lupus, Crohn's, JIA

Lupus

Initial presentation for abdominal pain and vomiting on 02/15/2024 and next 03/11/2024 and then for transport 03/12/2024
Malar rash, joint pain, ascites, rarely presents as Intestinal pseudo-obstruction due to vasculitis in abdominal wall, labs elevated ANA 1280,SSA, RNP and ds-DNA positive
Latent TB positive with quantiferon testing, fecal calprotectin positive, rifampin
Treated with solumedrol 1000 mg daily x 3 days then prednisone 60 mg daily

Crohn's disease

MG 10 yr with 4-7 loos bloody stools per day and during the night, MRI and scope positive for inflammatory changes in large and small intestines, treatment, then remission and reoccurence after 2 years off mes treated x 2 with prednisone, awaiting another biological

JIA(Juvenile idiopathic arthritis)

CJ:Patient presented at 5 yr with massively swollen knees, treated for years with methotrexate, stopped for 18 months, reoccurence at 16 years

Large effusion or synovitis. Subtle lucency with surrounding
sclerosis measuring 5 mm involving the medial femoral condyle.
Knee is otherwise normal. Osteochondritis Dessicans
Currently on Sulindac 200 mg bid, Ortho pending

Autoimmune thyroiditis

13 year old presented with depression, low energy and chronic nausea, poor appetite, initial Free T4 0.84, TSH 3.88, Thyroid peroxidase and thyroglobulin antibodies positive, back in school and working in childcare center

Case 2

36 week gestational age female 5#12 ounces, vaginal delivery, seen at 6 days of age, notified at 4 days of age high Succinylacetone on newborn screen, first cousin with Tyrosinemia 1,
Parents of both children, brothers married sisters,

Developing normally, growing normally

Tyrosinemia 1

Autosomal recessive
Commonly found in Saguenay-Lac-Saint Jean from Quebec and Finnish
Deficiency in Fumarylacetoacetate(FAH) with accumulation of substrates in the liver and kidney with a high rate of hepatocellular carcinoma
Tested on newborn screen for elevated succinylacetone(SA) on blood spot testingSA
Parents contacted directly and the baby was started on a special formula, Tyrex mixed with breast milk 50:50.
Treatment is restriction of protein and nitisinone, reduces the risk of early hepatocellular carcinoma. At risk for acute porphyria crisis eliminated with this medication
Monitoring with blood: comprehensive pane, cbc, AFP, SA levels. At risk for leukopenia, and thrombocytopenia every 6 months
If persistent liver failure, orthotopic liver transplant indicated
Annual MRI of liver and ophthalmology examinations
At risk for liver cancer or failure, renal failure and developmental delay

Case 3

LP-R She was noted at 10 months of age to have an enlarged liver below the right costal margin, initial Ast 27 and Alt 68, referred to the U of MI pediatric gastroenterology clinic and repeat labs were, 170, and 208 respectively, normal newborn screen, albumin and bili normal

FH positive for alpha-1-antitrypsin deficiency in great maternal grandmother, paternal grandmother and paternal uncle

Lab positive for alpha-1-antitrypsin deficiency Pizz phenotype
Follow up labs yearly including ultrasounds

Alpha-1-antitrypsin deficiency

Alpha-1-antitrypsin is a protease inhibitor(Pi) of the proteolytic enzyme elastase
Smoking and infection increase elastase burden in the lungs increasing degradation
Genetics autosomal co-dominant transmission, 1 in 80,000-100,000
Normal MM, alternatives S, z, Worst Pi*zz, and heterozygous some deficiency
AAT level below 80mg/dl at risk for emphysema
With severe deficiency, emphysema occurs at a younger age(46) and basilar hyperlucency
Spontaneous secondary pneumothorax
Homozygous recessive zz, bronchiectasis, develop adult onset hepatitis, cirrhosis, or hepatocellular carcinoma

Extrapulmonary manifestation panniculitis(hot, panful nodules to thigh or buttocks), vasculitis, IBD, fibromuscular dysplasia, and glomerulonephritis
Test if emphysema<45 yr, emphysema in nonsmoker, predominantly basilar emphysema, FH of emphysema and liver disease, adult onset asthma, unexplained chronic liver disease, panniculitis
40% significant liver disease, 90% emphysema if smokes, yearly FEV1

Case 4

AL First pneumonia at 3 months of age, chronic sinus and otitis with multiple tubes and adenoid removal
Status asthmaticus at 4 years with hospitalization, repeat pneumonia with hospitalization at 6 years, Severe GERD with Nissen fundoplication at 5 years
Eventually chronic RML pneumonia and ultimately now as a young adult, 26 year bronchiectasis, by 14 years with CT demonstrating scarring

FH half brother and sister with cystic fibrosis, she is a carrier
Diagnosis at Denver Children's Ciliary dyskinesia per electron microscopy

Current treatment, hyper sal nebs 7% twice daily, pulmozyme twice daily, Symbicort twice daily, vibration vest daily or twice with infection, sinus rinse twice daily, iatrogenic adrenal insufficiency
Subcutaneous Immunglobulin, Azithromycin mon, wed, Fri
Once a year hospitalized for clean out and bronchoscopy

Primary Ciliary Dyskinesia

1 in 7500 incidence
Autosomal recessive with 50 different genetic variants

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