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Hadju Cheney syndrome: Highest yield fact(s): Acroosteolysis hands and feet. Familial condition, autosomal dominant. Other: Short stature, craniofacial abnormalities, wormian bones, polycystic kidneys, neuro symptoms.

Caisson disease: Highest yield fact(s): Deep see divers rise too quickly and develop embolization of nitrogen gas causing infarction of brain, spinal cord and bones. Other: Spinal cord lesions more common than brain lesions. Treatment is hyperbaric oxygen therapy.

Riley-Day syndrome aka Familial Dysautonomia: Highest yield fact(s): Congenital neuropathy causing neuropathic joints. Autosomal recessive. Other: Disease has many other manifestations from superimposed autonomic nervous system, motor, and sensory abnormalities. See skeletal changes and chronic pneumonia on imaging. https://pubs.rsna.org/doi/10.1148/90.1.107

Behcet disease: Highest yield fact(s): Recurrent orogenital ulcers, uveitis. Look for GI ulcers and vascular abnormalities. Other: Can see dural sinus thrombosis, cerebral arterial aneurysm/occlusion/dissection, pulmonary aneurysms.
https://pubs.rsna.org/doi/10.1148/rg.e31

Still's disease aka juvenile idiopathic arthritis: Highest yield fact(s): most common pediatric chronic arthritis. Other: Large joints over small joints and cervical spine involvement. Look for erosions due to inflammatory arthritis. Can see pleural/pericardial effusions and hepatosplenomegaly.

Felty's syndrome: Highest yield fact(s): triad of rheumatoid arthritis, splenomegaly and neutropenia. Other: If see recurrent pulmonary infections in patient with RA, consider Felty syndrome (lung infection due to neutropenia).

Jaccoud's arthropathy: Highest yield fact(s): Deforming arthritis most classically developing following rheumatic fever or systemic lupus erythematosis. Ulnar subluxation of 2nd to 5th MCP joints that is non-erosive and reducible. Other: If erosions are present think rheumatoid arthritis.

Morquio syndrome: Highest yield fact(s): Autosomal recessive mucopolysaccharidosis. Presents early in life (2-3 years old). Causes dwarfism with many MSK abnormalities. Expect cervical spine imaging showing anterior vertebral beaking, platyspondyly, odonotoid hypoplasia and atlantoaxial subluxation. Other: May show pelvic radiograph with flared iliac wings. May have recurrent pulmonary infection.

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Pyknodysostosis: Highest yield fact(s): Autosomal recessive. **Osteosclerosis.** Dwarfism with micrognathia, straightening (obtuse angle) of mandible, finger agenesis that mimics acroosteolysis, underdeveloped mastoids and sinsuses, dense bones. Other: wormian bones, erosion distal clavicles.

Gaucher's disease: Highest yield fact(s): Inherited lysosomal storage disorder with glucocerebrosidase deficiency, causes disturbance of reticuloendothelial cells and histiocytes. Affects bone marrow, spleen and liver. Hepatosplenomegaly, iron overload, pancytopenia. Osteoporosis, lytic bone lesions, Erlenmeyer flask deformities, H-shaped vertebrae, osteonecrosis, pathological fractures. Other: See here for more info https://pubs.rsna.org/doi/10.1148/38.5.579.

Gardner's syndrome: Highest yield fact(s): Familial autosomal dominant GI polyposis, sinus, calvarial, and other MSK osteomas, gastric hamartomas. Other: Osteomas can entrap cranial nerves. Basically 100% risk of malignant transformation of colonic polyps over lifetime. Also associated with small bowel and pancreatic malignancies. Abdominal desmoid tumors.

Osteopoikilosis: Highest yield fact(s): Congenital hereditary condition with multiple bone islands (enostoses). Typically, incidental on imaging as bone is otherwise normal with no symptoms. An MSK don't touch legion. Admittedly not a multisystemic disease but I put it here as it is easily confused it with other multisystemic diseases (for example pyknodysostosis). Other: low signal on all MRI sequences.

Multicentric reticulohistiocytosis: Highest yield fact(s): Similar radiographically to gout and rheumatoid arthritis. Nodular swelling, distal and symmetric with erosions but no periarticular osteopenia and can have joint space widening. Other: this is a granulomatous disease primarily affecting joint synovial tissues and skin. Associated with breast and ovarian cancer.

Alkaptonuria (ochronosis): Highest yield fact(s): Autosomal recessive. Intervertebral disc calcifications from homogentisic acid deposition in nucleus pulposus. Osteopenia. Other: can have dark urine. Can also have joint space narrowing and chondrocalcinosis/subchondral sclerosis. CPPD can therefore present similarly on imaging but ochronosis is nucleus pulposis calcification and CPPD is annulus fibrosus calcification.

Menke's syndrome aka Kinky hair syndrome: Highest yield fact(s): X-linked recessive copper transport disorder. Kinky hair. Severe CNS involvement with highly tortuous intracranial arteries and brain parenchymal atrophy. Other: weak muscles, loose joints, occipital tendinous calcifications, osteoporosis

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Milkman's syndrome: Highest yield fact(s): Osteomalacia with Looser zone fractures, most common in lateral scapula, ribs, pubic rami and medial femoral neck. Highest yield fact(s): Note Looser zone stress fractures denotes osteomalacia NOT osteoporosis. Other: Can result from many multi-systemic processes including renal osteodystrophy, osteogenesis imperfecta, hyperthyroidism, etc.

Tuberous sclerosis: Highest yield fact(s): Clinical manifestations are cognitive impairment, seizures and adenoma sebaceum (reddish rash from angiofibromas on face). Radiology findings include cortical/subependymal tubers, subependymal hamartomas and giant cell astrocytomas, renal angiomyolipomas, cardiac rhabdomyomas. Other: White matter abnormalities with radial bands, association with polycystic kidney disease, lymphangioleiomyomatosis, pancreatic neuroendocrine tumors, hypopigmented ash leaf spots on skin, sclerotic bone lesions, MANY other findings. This is a phakomatosis. https://www.ajronline.org/doi/10.2214/AJR.13.12235

Caroli's disease: Highest yield fact(s): Autosomal recessive. Todani type 5 choledochal cyst with segmental cystic dilation of purely intrahepatic bile ducts. Look for central dot sign with central enhancement due to contrast in portal radicals surrounded by cystic bile ducts. Bile stasis can cause secondary pyogenic cholangitis and intrahepatic abscess, may present in young adulthood with right upper quadrant pain, fever, and jaundice. Other: High association with polycystic kidney disease (autosomal dominant and recessive) and medullary sponge kidney. https://www.ajronline.org/doi/full/10.2214/ajr.179.4.1791053

Typhlitis: Highest yield fact(s): Neutropenic colitis with marked cecum/TI thickening in setting of HIV/chemotherapy/immunosuppression from transplant, etc. If tested, they will give you clinical history to clue you towards presence of immunosuppression/malignancy such as leukemia/lymphoma. Other: High risk of perforation if perform colonoscopy or barium enema. This can have multisystemic imaging manifestations in terms of the primary disease process (lymphoma, organ transplant) and secondary typhlitis.

Castleman's disease aka giant lymph node hyperplasia: Highest yield fact(s): In adults 30s to 40s with typically large but benign mediastinal lymph adenopathy, adenopathy more common in chest than in abdomen/neck. HIV risk factor, more common in males. Other: Can be localized or systemic. If systemic may have hepatosplenomegaly and ascites.

Lymphangioleiomyomatosis (LAM): Highest yield fact(s): Cystic lung disease with thin-walled cysts uniformly throughout both lungs with often normal surrounding lung parenchyma. Association with tuberous sclerosis. Chylothorax and recurrent pneumothorax. Other: Renal angiomyolipomas, chylous ascites, cystic hygroma in neck, abdominal lymphadenopathy (Can be massive) and multifocal bone osteolysis. https://pubs.rsna.org/doi/10.1148/rg.253055006

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Rosai-Dorfman disease aka Sinus histiocytosis with massive lymphadenopathy. Highest yield fact(s): Often presents similar to lymphoma with lymphadenopathy and B symptoms. Cervical lymphadenopathy most common but can be seen throughout chest, abdomen, pelvis. Other: can have mass-like meningeal involvement, high uptake with FDG-PET, can have lung nodules, orbital involvement.

Erdheim Chester disease: Highest yield fact(s): Non-familial histiocytosis, systemic lipogranulomatous disease. Most commonly presents with MSK findings and bone pain with cortical thickening of long bones that spares the axial skeleton. Hairy kidney sign: bilateral perirenal soft tissue infiltration. Cystic lung disease. Other: Soft tissue infiltration can occur at many sites throughout body so many other findings possible. Can be retro-orbital, can cause diabetes insipidus due to pituitary infundibulum involvement, can have retroperitoneal fibrosis. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6047091/