

MI-COD

MSS INDIA- Case Of the Day



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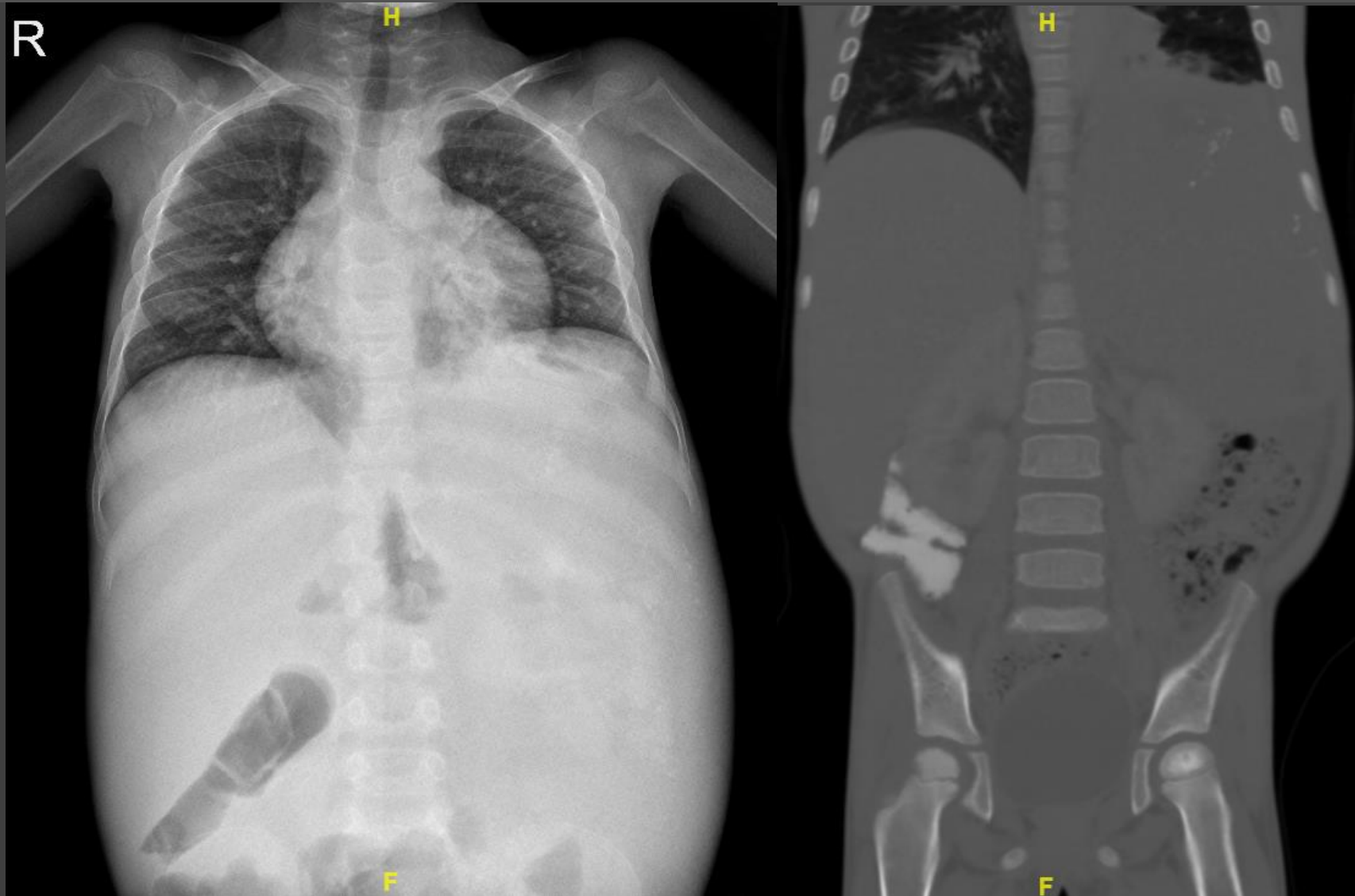
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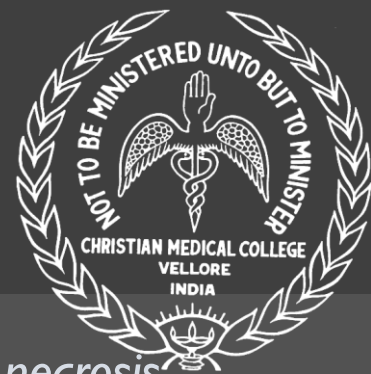
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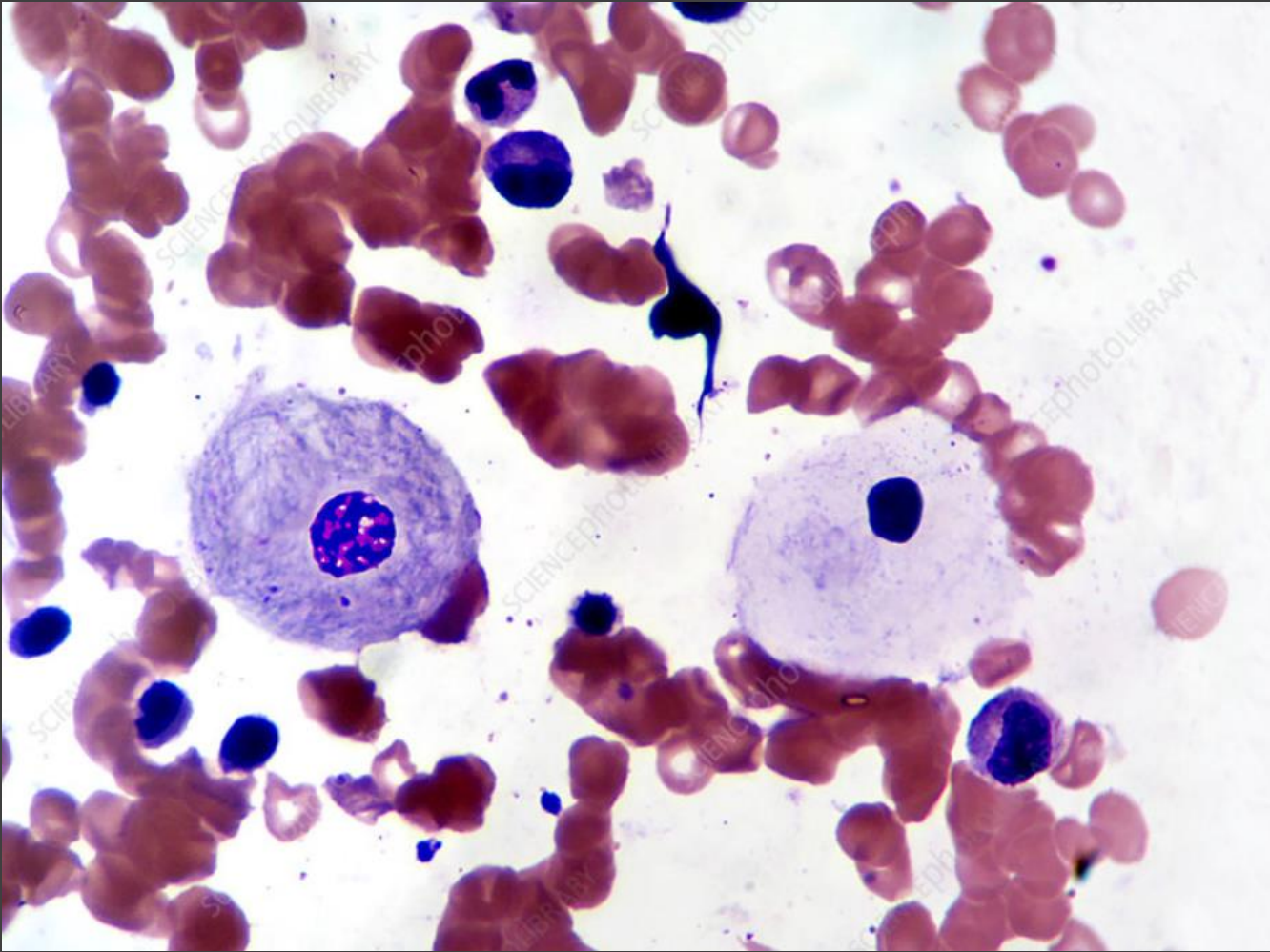
Gaucher's disease



Hepatosplenomegaly, widened metaphysis of proximal humeri, bilateral femoral head avascular necrosis



Gaucher's cell



Light micrograph showing Gaucher's cells, peripheral blood. Oil immersion technique. Magnification 600X when printed at 10 cm.

Gaucher cells in the macrophage-monocyte system typically found in the bone marrow with a **small nucleus, large cytoplasm** that have a characteristic **wrinkled-paper / onion layer like appearance**, resulting from intracytoplasmic substrate deposition

Gaucher's disease

The infographic features a central red silhouette of a human figure. Surrounding the figure are six icons representing different body parts: an eye, a brain, a blood drop, lungs, a bone, and a liver/spleen. Each icon is accompanied by a heading and a list of symptoms. The layout is clean and uses a color palette of red, blue, and white.

ORD
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Gaucher Disease

Signs and Symptoms

EYES
Difficulties in eye movement, yellow spots in eyes

BRAIN
Difficulties in coordination and motor skills, cognitive problems, seizures, developmental delays

BLOOD
Anemia, fatigue, thrombocytopenia

LUNGS
Breathing difficulties

BONES
Joint pain and damage, weakened bones, arthritis, osteoporosis, osteonecrosis

LIVER AND SPLEEN
Liver and spleen enlargement

For more information, visit:
www.hopkinsmedicine.org
my.clevelandclinic.org
www.mayoclinic.org

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- Prototypical lysosomal storage disease due to deficiency of the enzyme glucocerebrosidase
- Accumulation of glycolipids in the reticuloendothelial system leads to anemia, thrombocytopenia, and hepatosplenomegaly
- Skeletal manifestations due to build up of the glycolipids in the bone marrow and lead to the most debilitating aspects of GD.
- Treatment results in improvement of marrow infiltration however it takes much longer than the visceral and hematologic manifestations.
- Imaging plays a key role in both initial diagnosis and treatment monitoring. MRI of the abdomen is used to monitor liver and spleen volumes. MRI of the femora and lumbar spine is used for evaluation of bone marrow infiltration burden.