

FABRY, GAUCHER, INCIDENTALOMAS, AND MORE!

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Bohm P, et al. Adult Gaucher disease in association with primary malignant bone tumors. Cancer. 2001;91(3):457-62.
Grumbach MM, et al. Management of the clinically inapparent adrenal mass. Ann Intern Med. 2003;138(5):424-9.
Paladini G, Sala PG. Anion gap in multiple myeloma. Acta Haematol. 1979;62(3):148-52.

Take home points:

1. Fabry's disease = lysosomal storage disease → accumulation in cells → microvascular obstruction → LVH, diastolic dysfunction, MI, ESRD, strokes
2. Gaucher's disease = accumulation of beta-glucocerebrosides in reticuloendothelial cells, causing cytopenia, hepatosplenomegaly, pathologic fractures.
3. Anion gap in multiple myeloma: decreased gap in IgG myeloma, normal/increased gap in IgA myeloma

What is Fabry's disease?

- X-linked, recessive lysosomal storage disease due to deficiency of alpha-galactosidase A
- Classic phenotype have undetectable levels of the alpha-galactosidase A; there are many patients with partial activity of alpha-galactosidase and therefore milder disease
- The disease is panethnic, and affects 1:50,000 males; many cases are not diagnosed until young adulthood
- Look for cardiac (LVH, diastolic dysfunction, MI), renal (proteinuria, ESRD), and cerebrovascular manifestations. Fabry's disease is an underappreciated cause of "idiopathic" LVH. Organ dysfunction is caused by microvascular obstruction from engorged vascular endothelial cells.
- Enzyme replacement therapy (and potential cure) is now available

What is Gaucher's disease?

- Several types – remember *type I* (most common variant, can present in adulthood)
- Prevalent in Ashkenazi Jewish population
- Deficient enzyme causes accumulation of beta-glucocerebrosides in reticuloendothelial cells, causing cytopenias, hepatosplenomegaly, pathologic fractures.
- Can mimic infiltrative diseases
- Enzyme replacement therapy is now available (replaces bone marrow transplantation)

Anion gap in multiple myeloma:

- IgG is a cation; therefore, in IgG myeloma, the anion gap is *decreased*
- IgA is an anion; therefore, in IgA myeloma, the anion gap is *normal or increased*
- You can follow the anion gap (to look for normalization) in the treatment of myeloma

What do I do with an adrenal incidentaloma?