Presentation of COPA Syndrome in an Adult: A Newly Described Primary Immunodeficiency

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Presentation of COPA Syndrome in an Adult: A Newly Described Primary Immunodeficiency

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BACKGROUND

- COPA syndrome is a newly discovered hereditary immune dysregulatory primary immunodeficiency which is autosomal dominant with variable penetrance.[1,2]
- The mutated gene, COPA, encodes the alpha subunit of the coatomer complex-I which is involved in the transit of proteins between the Golgi complex and endoplasmic reticulum.[1–3]
- Most commonly presents before the age of 5 with a female predominance.[1,3]
- Early symptoms include shortness of breath, cough, and joint pain.[1,2]

CLINICAL FEATURES	PREVALENCE
Pulmonary Hemorrhage	High
Interstitial Lung Disease	Moderate
Renal Disease	Very High
Arthritis	High
Skin Disease	Rare

CASE DESCRIPTION

55-year-old female with a past medical history of chronic cavitary aspergillosis status post three lung resections, pulmonary hemorrhage, undifferentiated connective tissue disease, adrenal insufficiency, and recurrent squamous cell carcinoma presented for evaluation of immunodeficiency due to unusual and recurrent pulmonary infections (aspergillosis and mycoplasma). Physical exam was remarkable for a left forehead scar and nose lesion being worked up for squamous cell cancer. Her significant medications included posaconazole and previously voriconazole.

RESULTS

- Negative: Rheumatoid factor (RF), cyclic citrullinated peptide (CCP), anti-nuclear antibodies, anti-myeloperoxidase (MPO) and anti-proteinase 3 (PR3) antibodies.
- Buccal Biospy: Negative for Sjogren's Disease.
- CT Chest: multiple thick-walled cystic lesions consistent with aspergillosis and scattered ground glass opacities bilaterally.
- COPA Gene: Positive in July 2016.

DISCUSSION

- Our case highlights the findings of recurrent cavitary aspergillosis and recurrent skin cancers in COPA syndrome.
- COPA syndrome may be more common than other primary immunodeficiencies due to being autosomal dominant.[1]
- While COPA syndrome should always been considered in young patients with pulmonary disease, it is also important to recognize different presentations and consider undiagnosed COPA syndrome in adults with suspected immunodeficiency.[2]
- When suspected, patients should be tested for COPA gene mutation to allow for further development of diagnostic criteria and treatment regimens.

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