14 Year Old Boy With ill-defined Immunodeficiency

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RATIONALE: Common Variable Immune Deficiency (CVID) is characterized by recurrent infections, reduced IgG, low levels of IgA and/or IgM, impaired antibody response and normal or mildly abnormal cellular immunity. Noncaseating granulomas are commonly seen in patients with CVID and can be misdiagnosed as sarcoidosis.

METHODS: 14 year old boy presented at age 2 with recurrent infections, chronic diarrhea and family history of a sister with vague rheumatological complaints who died of staphylococcus aureus sepsis and lung abscesses at age 5.

RESULTS: Evaluation revealed lymphopenia, IgG2 and IgG4 deficiency, and polysaccharide functional antibody deficiency. Despite IVIG, he continued to have recurrent infections and failure-to-thrive. Recently he presented with a rapidly enlarging palatal fistula. Biopsies of skin and sinuses showed noncaseating granulomas. This prompted a reassessment of his diagnosis. Evaluation off of IVIG for 4 months revealed lymphopenia (ALC: 1003/mm3), low IgG2 (<17 mg/ml) and IgG4 (<4 mg/ml) and no post-immunization response for tetanus and pneumococcus. Total IgG

and IgA were normal and IgM was elevated (579 mg/dL). ANCA, ANA, NBT, DHR, HBV, HCV, HIV, ACE and sweat test were normal. Chest CT showed bronchiectasis and persistent hilar adenopathy. Possible diagnoses included CVID, sarcoid, malignancy, IPEX, Wegener's granulomatosis and lethal midline granulomatous disease. The patient is currently receiving methotrexate, infliximab and prednisone to treat his granulomatous disease.

CONCLUSIONS: We present an interesting patient whose clinic and laboratory results are a diagnostic puzzle. Our patient does not fulfill criteria for any known immunodeficiency