STAT-3 DEFICIENT CHILD FOUND TO BE SYMPTOMATIC CARRIER OF CYSTIC FIBROSIS
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Introduction: Although unlikely, the presence of one immune deficiency does not exclude another. We present a child with STAT-3 deficiency and Hyper IgE Syndrome (HIES) who developed pseudomonas pneumonia and was also found to be a symptomatic carrier for cystic fibrosis (CF).
Case Description: A 7-year-old girl with STAT3 deficiency, hyper IgE syndrome and bronchiectasis was diagnosed at the age of 3 and had multiple pneumonias with staphylococcus and pseudomonococ. She suffers from severe eczematous rash and has bony abnormalities and dentition abnormalities. Her newborn screening for CF was negative and a sweat test for CF during her initial work-up was below cut-off. A recent flare of her bronchiectasis was not responding to treatment for staphylococcus, and fungal treatment was added while awaiting sputum culture. The sputum culture was positive for Pseudomonas aeruginosa (PA). Whole exome sequencing for CF was performed and she was discovered to be heterozygous for a known pathogenic variant. She was treated with appropriate antibiotics and started on a prophylaxis regimen of inhaled antibiotic for CF.
Discussion: HIES usually renders the patient susceptible to staphylococcus and sometimes aspergillus. It is not associated with Pseudomonas aeruginosa infection. There have been multiple reports of symptomatic carriers of Cystic Fibrosis. Just as a high index of suspicion is necessary to diagnose primary immune deficiencies, a high index of suspicion is needed in caring for these patients.