Chronic cough in a 5 year-old girl: putting the puzzle together

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A 5-year-old girl was referred to the Pulmonology Division in our hospital due to uncontrolled asthma. She had a past medical history of a chromosomal mosaicism, mild global developmental delay, dyskinesias and a posture defect with flat valgus foot. At one year of age, she presented with recurrent episodes of wet cough, increased work of breathing, dyspnea on exertion and wheezing. She was diagnosed with asthma by her pediatrician and began treatment with ICS and montelukast with no improvement of her symptoms. In addition, she had multiple consultations to the ER and one hospital admission due to worsening symptoms, where she received several courses of oral steroids and antibiotics. When evaluating for other comorbidities, the patient showed some feeding difficulties but no history of failure to thrive or chronic diarrhea.

On physical examination, the patient had a good general appearance and was well nourished (weight 18kg, height 108.5cm, both in 50th percentile for age) with a SpO2 93% in room air. ENT exploration was normal, with no nasal discharge or polyps. No stridor was present and mild intermittent intercostal retractions were noted. Lung auscultation revealed bilateral diminished breath sounds and diffuse wheezing and crackles. Cardiovascular and abdominal examinations had no abnormal findings. Extremities did not show cyanosis or digital clubbing.

Given her history and physical examination, a complete work up for chronic suppurative lung disease was performed: CBC, electrolytes, renal function and liver function were normal. Chest X-Ray was reported as normal. She had a high (undeterminate) sweat test with a normal control. An immune work out showed an undetectable IgE, normal IgA and IgM, but low IgG. ELISA test for HIV was negative. Barium swallow study described a normal anatomy, no gastroesophageal reflux, but presented cricopharyngeal incoordination and broncoaspiration during the study.

Bronchoscopy documented thick purulent tracheobronchitis and normal airway anatomy. BAL cultures were positive for *P. aeruginosa* and cytology showed abundant lipid-laden macrophages. The patient was admitted for IV antibiotics and was discharged with nasogastric tube feedings.

Her chest CT scan revealed a bilateral centrilobular micronodules, incipient bronchial dilatation, with no other abnormal findings reported.

She continued with persistent productive cough therefore other studies were completed for immunodeficiency. NBT test was normal. Flow cytometry test documented CD4+ T lymphopenia and B lymphopenia with elevated levels of NK cells. IgG levels remained low and alpha fetoprotein levels were high. On follow up appointments an ataxic gait was noted. The diagnosis of ataxia telangiectasia (AT) was made and confirmed by genetic analysis. She was started on IGIV. After several months, her lung condition worsened, and another BAL culture resulted positive for *P. aeruginosa*. We decided to initiate an eradication antibiotic protocol for 14 days and was discharged with 8 weeks of nebulized Tobramycin, 4

weeks of oral Ciprofloxacin, with oral Clarithromycin 3 days/week during the entire protocol. Once the scheme was completed, a new BAL culture was taken and it resulted negative for *P.aeuruginosa.*

The patient has remained stable from a respiratory standpoint with no exacerbations or hospitalizations during the past 2 years.

Ataxia telangiectasia is a rare autosomal recessive disorder with a broad range of clinical manifestations and different forms of presentation. Lung involvement is a common feature and patients may present with early recurrent respiratory symptoms that could resemble other conditions, making the diagnosis a real challenge. Chronic lung disease is a major comorbidity and there is lack of evidence on how to prevent and treat this condition. There is a need for consensus on treatment protocols to improve lung health in these patients.

References

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