CASE PRESENTATION

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A 15 year-old teenage girl, with multiple autoimmune manifestations and hypogammaglobulinemia.

FAMILY HISTORY

- Non-consanguineous parents.
- 26 year-old brother with progressive muscular dystrophy (wheelchair). Healthy 20 year-old brother.
- Paternal and maternal aunts with rheumatoid arthritis.
- Great uncle with vitiligo.

PAST MEDICAL HISTORY

- First manifestation was at the age of 3 years, when she presented with **alopecia areata**.
- Months later she was diagnosed with **atopic dermatitis**, and hypocromic spots were noted, diagnosed as **vitiligo**.
- **Food allergy**: cow milk, soy and egg. Also, allergy-like symptoms (persistent rhinitis) during early childhood, now asymptomatic.
- Recurrent upper **respiratory tract** infections starting at 2 years of age. Three pneumoniae, last one in 2014.

PAST MEDICAL HISTORY

- She was 6 year-old when developed **autoimmune hypothyroidism**, and began replacement treatment.
- 9 year-old: bilateral ptosis and right exotropia, was diagnosed as **myasthenia gravis**, with spontaneous recovery. During the same year, she presented with **localized scleroderma**.

PHYSICAL EXAM

- Alopecia patches.
- Generalized hypo-pigmented skin patches alternating with normal pigmentation areas.
- Two circular, indurated and hyperpigmented plaques of about 5cm, localized over the chest and interscapular areas.
- Normal cardiopulmonary and abdominal findings.

LAB WORKUP

CBC	
WBC	9700mm^3
Neutrophils	7200mm^3
Lymphocytes	1200mm^3
Monocytes	700mm^3
Platelets	282,000mm^3

Autoantibodies	
Anti-RNP	<2
Anti-SM	<2
Anti-Jo-1	<2
ANA	negative
Anti- ENA6	negative

Immunological Assessment	
IgA	71.1 mg/dL
IgG	461 mg/dL
IgM	48.8 mg/dL
IgE	<1mg/dL
CD3+	86% 1188
CD4+	63% 749
CD8+	33% 402
CD19+	Not detected
CD16+CD56+	16% 222

Anti-thyroid autoantibodies	
Anti-thyroglobulin (TG)	180
Anti- thyroid peroxidase (TPO)	>1000

Complement	
C3	125
C4	24.5

Normal liver and kidney function tests

CLINICAL COURSE

- Developed **muscular weakness** and lower limb motor impairment, suspected autoimmune myopathy; evoked potential tests reported a non-specific disorder in a wave-dispersion pattern, of spontaneous remission. Normal MRI. (No biopsy)

- Mild thoracolumbar **scoliosis** diagnosed, treated conservatively.

- **Chronic diarrhea,** no isolates, no other symptoms, considered inflammatory bowel disease. No colonoscopy.

- In recent months with **progressive dyspnea**, poor exercise tolerance, peripheral oxygen desaturation, and oxygen dependency during sleep.

    - **HIGH RESOLUTION COMPUTED TOMOGRAPHY:** Basal cylindrical **bronchiectasis**.
    - **LUNG FUNCTION TESTS :** Reduced rate of carbon monoxide uptake and **restrictive** ventilatory pattern.