A 15 year old female with leukopenia

Ahmad Hamad, MD
Allergy & Immunology Fellow
University of North Carolina, Chapel Hill, NC

Thomas A.Fleisher, MD
Chief, Department of Laboratory Medicine
NIH Clinical Center

History

HPI:

- 15 yo female.
- Admitted for dyspnea. Diagnosed with pulmonary hypertension.

PMH & PSH:

- Congenital deafness.
- BOR syndrome.
- Pneumonia at age 9.
- UTI at age 13.

FH:

- No history of PID.
- Mother and brother have BOR syndrome.

Chart review:

- History of leukopenia: unclear etiology per heme, possible viral suppression during pervious visits.
- BM bx was done twice.
- No history of recurrent infections or failure to thrive.
- Persistent elevation of ESR and CRP with negative infectious and rheumatologic workup.

Labs

CBC with diff	Age 12	Age 13	Age 14	Age 15
WBC (4.5 -11)	2.2	1.4	3.1	2
HGB (12 - 16)	7.6	4.7	11.9	8
PLTS (150-440)	307	249	237	100
ANC (2-7.5)	1.4	0.7	1.8	1.5
ALC (1.5 -5)	0.8	0.5	1.2	0.4
Abs monocytes (0.2-0.8)	0	0	0	0
Abs Eos (0- 0.4)	0	0	0	0
Abs Baso (0-0.1)	0	0	0	0

Lymphocyte count	
CD3% 61-86	89 (H)
Absolute CD3 count 915-3400	844 (L)
CD4% (34-58)	53
Absolute CD4 count 510-2320	502 (L)
CD8% 12-38	35
Absolute CD8 count 180-1520	332
CD:CD8 ratio 0.9-4.8	1.5
CD 19% (B cells) 7-23	<1 (L)
Absolute CD19 count 105-920	< 10 (L)
CD 16/56% NK cells 1-27	10
Absolute CD16/56 15-1080	95

Bone marrow biopsy	
Age 12	Age 13
Normocellular bone marrow (85%) with erythroid predominant trilineage hematopoiesis and absent iron stores.	Hypocellular bone marrow (70%) with erythroid predominant trilineage hematopoiesis and absent iron stores
Routine cytogenetic results reveal a normal karyotype	Routine cytogenetic results are normal

Immunoglobulins	Age 15
ESR (0-20)	66(H)
IgG (600-1700)	3701(H)
IgM (35-290)	93
IgA (40-400)	228
Tetanus IgG	2.95
Diphetheria IgG	0.66
Total IgE	< 2

Hospital course and follow up

- Diagnosed with pulmonary hypertension WHO I : started on sildenafil and inhaled prostacyclin.
- Possible undifferentiated connective tissue disease: started on high dose steroids and later on a steroid sparing agent.
- Persistent leukopenia, B cell lymphopenia and monocytopenia.
- Shortly after diagnosis, she was hospitalized for pneumonia.

GATA2 genetic analysis:

Diagnosis:

Gata2 genetic analysis revealed heterozygosity for a sequence variant defined as c.1128 C>A that is predicted to result in premature protein termination (p. Tyr376) : not previously reported.

Treatment :

Bone marrow transplantation.

Discussion:

Spectrum of clinical symptoms.

Clues to make a diagnosis.

Management of disease.

Thank you