

Worsening lung findings in a SCID patient

Melanie A. Ruffner, MD, PhD
 Fellow, Division of Allergy and Immunology
 The Children's Hospital of Philadelphia



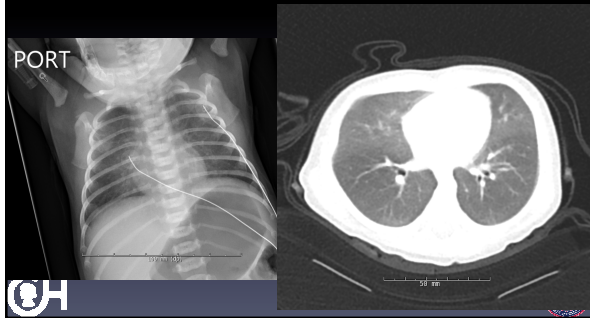
Case Background

- 2 wk full term female infant
- No pregnancy or delivery complications
- Significant family history of immunodeficiency
- NJ NBS positive

% CD3=12 (Absolute CD3=3)
 %CD4=10 (Absolute CD4=3)
 %CD8=7 (Absolute CD8=2)
 %NK=73 (Absolute NK=18)
 %CD19 (Absolute CD19=2)

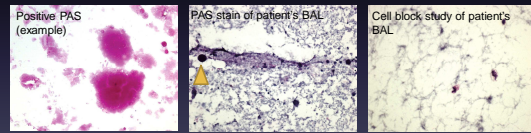


"Chesty Cough"



Initial Admission Course

- Developed progressive coarse rales and brief desaturations
- LDH fell to 1105
- Bronchoscopy with BAL was performed with 470 RBC, 270 WBC, 65% neutrophils, 16% monocytes and 10% lymphocytes.
- PAS stain was negative
- Complete infectious workup was negative



Despite this, she worsened significantly with deteriorating respiratory status with fever on day 7 of admission

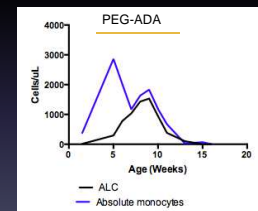
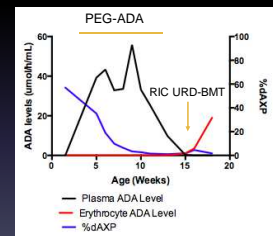


Patient/Date	hemolysate nmol/h/mg		PBL nmol/h/mg		µmol/ml RBC		
	ADA	PNP	ADA	PNP	AXP	dAXP	% dAXP
	0.0	1180			0.744	0.985	57.0
Normal levels	63.0 ± 41.4	1338 ± 441	1197 ± 516	3007 ± 824	1.465 ± 0.38	<0.00 2	<0.2

ADA, adenosine deaminase; PNP, purine nucleoside phosphorylase
 AXP (dAXP)=total adenosine (deoxyadenosine) nucleotides.
 % dAXP= (dAXP/AXP+dAXP) x 100



Management with PEG-ADA



Followup

- Labs from 16 months of age (13 months post-transplant):
 - Engraftment is 100%
 - Absolute CD3+ 953, NK cell numbers are normal, Absolute CD19+ normal 546 with 8% switched memory B cells. Transitioned off IVIG, 1 yr post-transplant all immunoglobulin levels are normal
 - PHA 39% of control
 - TRECs slowly improving and have normalized, and oligoclonality on spectratyping has been progressively decreasing

At 6 months (shortly after hospital discharge) she was referred to early intervention due to speech and motor delay. Currently at 16 months sitting and cruising but cannot walk independently.

