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Esosa Adah

*Children's Mercy Kansas City*

Nikita Raje

*Children's Mercy Hospital*

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# ADA Deficiency: Improvement in Immune System after Enzyme Replacement Therapy

Esosa Adah MD, Nikita Rajc MD, MSc

Division of Allergy, Immunology, Pulmonary and Sleep Medicine, Department of Allergy and Immunology, Children's Mercy Kansas City

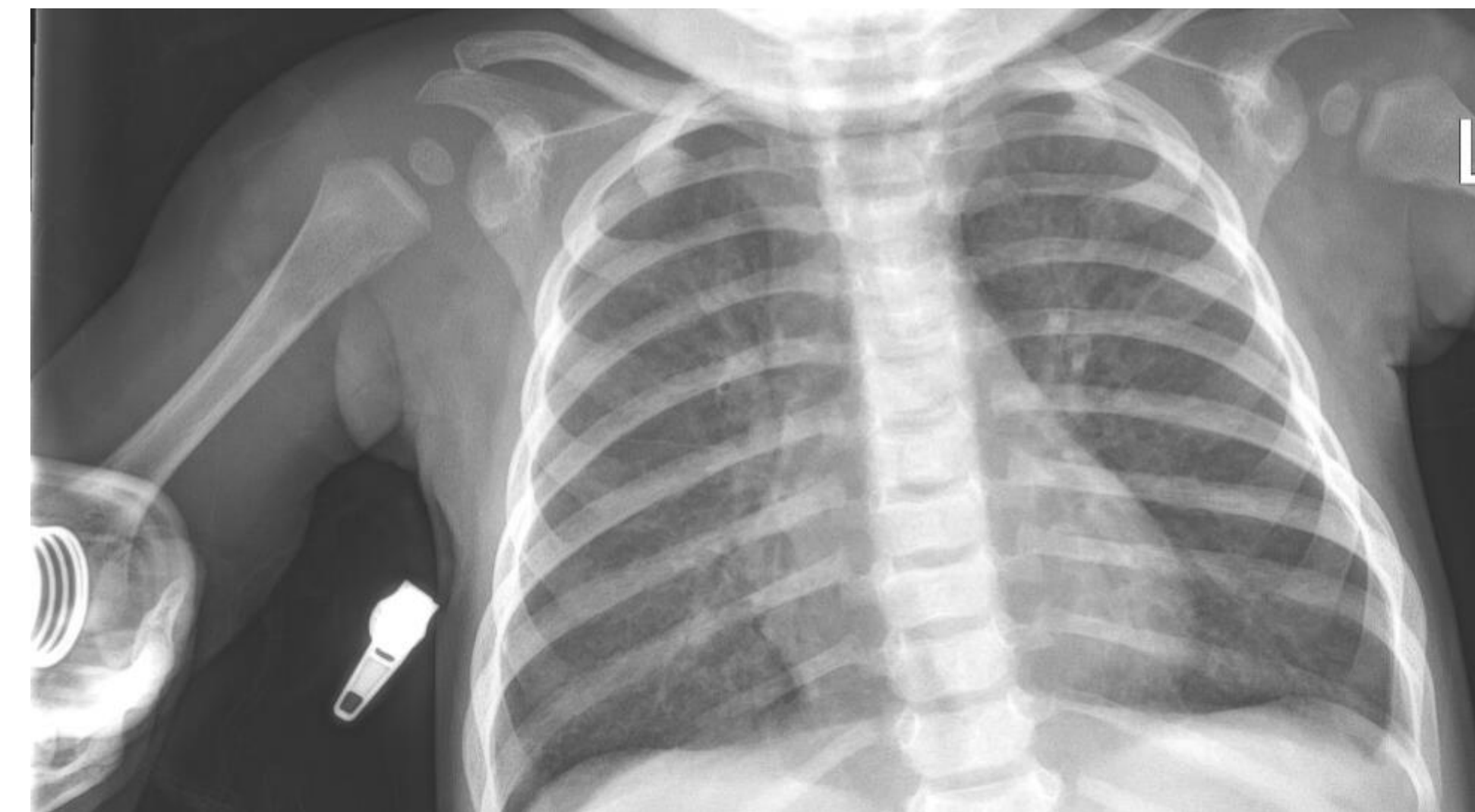
## Introduction

- Adenosine deaminase (ADA)-deficient severe combined immunodeficiency (SCID) is characterized by severe lymphocytopenia affecting T-, and B- lymphocytes, with non-immunological manifestations including neurodevelopmental deficits, sensorineural deafness, and skeletal abnormalities
- Treatment modalities include enzyme replacement therapy(ERT) and/or autologous gene therapy or hematopoietic stem cell treatment
- We present a case of a 9 month old female with ADA-deficient SCID who underwent enzyme replacement therapy prior to stem cell transplant

## Patient Presentation

- A 9-month-old female with a history of failure to thrive, hypotonia, chronic cough and loss of developmental milestones had genetic testing done that showed variants ADA c.911 T>G, c.302G>A
- She was admitted for further workup
- Patient diagnosed with T-B-NK+ ADA deficiency SCID after lymphocyte cytometry and ADA levels obtained

## Imaging



- Figure 1
- Table 1
- Table 2

## Lab Results

	Presentation	2 weeks after ERT	2 months after ERT
Total T cells (CD3+)mm <sup>3</sup>	117 (49%)	516 (51%)	1241 (47%)
CD4 cells mm <sup>3</sup>	55 (23%)	293 (29%)	792 (30%)
CD8 cells mm <sup>3</sup>	50 (21%)	222 (22%)	423 (16%)
CD19 cells mm <sup>3</sup>	7 (3%)	162 (16%)	766 (29%)
NK cells mm <sup>3</sup>	108 (45%)	324 (32%)	607 (23%)

Table 1

Weeks on PegADA	Plasma ADA Umol/h/ml	RBC Nucleotides Umol/ml RBC dAXP (%dAXP)
0	Undetectable	0.299(14.7)
2	134.7	0.130(7.9)
6	212.6	0.028(1.6)
15	153	0.003(0.2)
20	147.4	0.002 (0.1)

Table 2

## Figures and Tables

- Figure 1 shows initial chest x-ray showing pulmonary opacities in our patient
- Table 1 shows the lymphocyte progression after Elapegamase was initiated
- Table 2 shows ADA and dAXP levels in response to PegADA

## Discussion

- ADA deficiency SCID is known to present with T-B-NK-immune phenotype; however, our patient presented with T-B-NK+ cells
- Our patient showed clinical and immunological improvement from ERT
- Our patient is now s/p stem cell transplant after 20 weeks of ERT
- ERT should be considered in ADA SCID patients while awaiting definitive curative treatment

## References

- Secord E, Hartog NL. Review of Treatment for Adenosine Deaminase Deficiency (ADA) Severe Combined Immunodeficiency (SCID). *Ther Clin Risk Manag.* 2022 Sep 22;18:939-944. doi: 10.2147/TCRM.S350762. PMID: 36172599; PMCID: PMC9512634.
- Hershfield M. Adenosine Deaminase Deficiency. 2006 Oct 3 [updated 2017 Mar 16]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2023. PMID: 20301656.