

Deficiency of [adenosine deaminase 2](#) is characterized by vasculitis, early-onset strokes, immunodeficiency, and bone marrow failure. We describe a novel pathogenic mutation affecting a consensus N-linked glycosylation sequence and illustrate the essential role of glycosylation in the biology of ADA2 ¹⁾.

¹⁾

Lee PY, Huang Y, Zhou Q, Schnappauf O, Hershfield MS, Li Y, Ganson NJ, Sampaio Moura N, Delmonte OM, Stone SS, Rivkin MJ, Pai SY, Lyons T, Sundel RP, Hsu VW, Notarangelo LD, Aksentijevich I, Nigrovic PA. Disrupted N-linked glycosylation as a disease mechanism in deficiency of ADA2. *J Allergy Clin Immunol*. 2018 Jun 21. pii: S0091-6749(18)30902-3. doi: 10.1016/j.jaci.2018.05.038. [Epub ahead of print] PubMed PMID: 29936104.

From:

<https://neurosurgerywiki.com/wiki/> - **Neurosurgery Wiki**

Permanent link:

https://neurosurgerywiki.com/wiki/doku.php?id=adenosine_deaminase_2

Last update: **2024/06/07 02:49**

