Novel mutation in CECR1 gene associated with deficiency of adenosine deaminase -2 presenting as severe congenital neutropenia

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Abstract

Children with congenital neutropenia frequently require hospitalization due to febrile neutropenia. Deficiency of Adenosine DeAminase -2(DADA-2) an autosomal recessive disorder caused by a mutation in CECR1 gene. It is an autoinflammatory disease presenting with autoimmunity and features of immunodeficiency. It usually presents in early childhood with recurrent stroke and vasculitis features. Here, we report a young male with CECR1 mutation presenting predominantly with neutropenia.

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`DADA -12.6.22.docx` available at https://authorea.com/users/485405/articles/572746-novel-mutation-in-cecr1-gene-associated-with-deficiency-of-adenosine-deaminase-2-presenting-as-severe-congenital-neutropenia