

Open Peer Review on Qeios

Erythrocyte Adenylate Kinase Deficiency

National Cancer Institute

Source

National Cancer Institute. <u>Erythrocyte Adenylate Kinase Deficiency</u>. NCI Thesaurus. Code C131640.

An autosomal recessive disorder caused by mutation of the AK1 gene. It is associated with moderate to severe non-spherocytic hemolytic anemia and, in some cases, with intellectual disability and psychomotor impairment.

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