

Genetic form of anemia defined molecularly

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Sideroblastic anemia is a form of anemia caused by an inability to incorporate iron into hemoglobin, something that is essential if the molecule is to perform its vital function of carrying oxygen from the lungs to the tissues.

Recently, a patient with sideroblastic anemia was found to have a mutation in their GLRX5 gene that led to GLRX5 protein deficiency.

Tracey Rouault and colleagues, at the National Institute of Child Health and Human Development, Bethesda, have now provided insight into how GLRX5 protein deficiency causes sideroblastic [anemia](#).

Specifically, they find that in human cells, GLRX5 is essential for generating iron-sulfur clusters (molecular groups that facilitate a wide range of cellular activities, including sensing of iron and [oxygen](#)) and maintaining normal levels of iron in cellular compartments known as mitochondria and the cytosol.

Further analysis revealed a molecular explanation for why GLRX5 protein deficiency caused disease in only one cell type in the body, the red blood cell.

More information: View this article at:
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