

Children's Mercy Kansas City

SHARE @ Children's Mercy

Manuscripts, Articles, Book Chapters and Other Papers

1-4-2018

Cabot rings and marked anisopoikilocytosis in Imerslund-Gräsbeck syndrome.

Derrick L. Goubeaux
Children's Mercy Hospital

Weijie Li
Children's Mercy Hospital

Follow this and additional works at: <https://scholarlyexchange.childrensmercy.org/papers>



Part of the [Hematology Commons](#), and the [Pathology Commons](#)

Recommended Citation

Goubeaux DL, Li W. Cabot rings and marked anisopoikilocytosis in Imerslund-Gräsbeck syndrome. *Blood*. 2018;131(1):153. doi:10.1182/blood-2017-10-809178

This Article is brought to you for free and open access by SHARE @ Children's Mercy. It has been accepted for inclusion in Manuscripts, Articles, Book Chapters and Other Papers by an authorized administrator of SHARE @ Children's Mercy. For more information, please contact hlsteel@cmh.edu.



Cabot rings and marked anisopoikilocytosis in Imerlund-Gräsbeck syndrome

Derrick L. Goubeaux and Weijie Li, Children's Mercy Hospital



A 12-year-old female presented with fatigue and weight loss. She appeared pale and had gray streaks in her hair. She had a history of chronic anemia, and some family members also had anemia. Laboratory tests showed macrocytic anemia (hemoglobin 5.2 g/dL, mean corpuscular volume 107.3 fL) and thrombocytopenia (platelet count $129 \times 10^9/L$), increased blood homocysteine (111.6 $\mu\text{mol/L}$, reference range 4.7-10.3), and decreased vitamin B₁₂ (111 pg/mL, reference range 260-935). Wright-stained peripheral blood smear showed macrocytosis, anisopoikilocytosis, a few polychromatophilic red blood cells with rare purple Cabot rings (panel A, arrow; original magnification $\times 1000$), and coarse basophilic stippling (panels B and C; original magnification $\times 1000$). Workup for vitamin B₁₂ deficiency identified

a homozygous pathogenic variant in the *AMN* gene, which is consistent with Imerlund-Gräsbeck syndrome (IGS), a rare autosomal recessive disorder caused by a defect in the receptor of vitamin B₁₂-intrinsic factor complex in terminal ileum. This receptor is composed of 2 proteins, amnionless (*AMN*) and cubilin (*CUBN*). Mutations of either the *AMN* or *CUBN* gene can cause IGS.

Cabot rings are thin, thread-like ring- or "figure eight"-shaped red blood cell inclusions, likely remnants from mitotic spindles. They are rarely seen in peripheral blood, and their presence indicates a defect in erythrocyte production, especially in pernicious anemia and lead poisoning.



For additional images, visit the ASH Image Bank, a reference and teaching tool that is continually updated with new atlas and case study images. For more information, visit <http://imagebank.hematology.org>.



blood[®]

2018 131: 153

doi:10.1182/blood-2017-10-809178

Cabot rings and marked anisopoikilocytosis in Imlerslund-Gräsbeck syndrome

Derrick L. Goubeaux and Weijie Li

Updated information and services can be found at:

<http://www.bloodjournal.org/content/131/1/153.full.html>

Articles on similar topics can be found in the following Blood collections

[BloodWork](#) (688 articles)

[Free Research Articles](#) (5369 articles)

[Red Cells, Iron, and Erythropoiesis](#) (945 articles)

Information about reproducing this article in parts or in its entirety may be found online at:

http://www.bloodjournal.org/site/misc/rights.xhtml#repub_requests

Information about ordering reprints may be found online at:

<http://www.bloodjournal.org/site/misc/rights.xhtml#reprints>

Information about subscriptions and ASH membership may be found online at:

<http://www.bloodjournal.org/site/subscriptions/index.xhtml>