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](https://scholarlyexchange.childrensmercy.org/papers)

Part of the Hematology Commons, and the Pathology Commons

**Recommended Citation**


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 library@cmh.edu.
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 × 10⁹/L), increased blood homocysteine (111.6 μmol/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 ×1000), and coarse basophilic stippling (panels B and C; original magnification ×1000). Workup for vitamin B₁₂ deficiency identified a homozygous pathogenic variant in the AMN gene, which is consistent with Imerslund-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.
Cabot rings and marked anisopoikilocytosis in Imerslund-Gräsbeck syndrome

Derrick L. Goubeaux and Weijie Li