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Cabot rings and marked anisopoikilocytosis in Imerlund-Gräsbeck syndrome

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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.



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