# A Young Adult with Aplastic Anemia and Gray Hair

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## CASE

A man in his early twenties presented with hematuria and moderate pancytopenia [white blood cell count, 1.9 X $10^9$/L (reference, 4.2 X $10^9$/L to 9.9 X $10^9$/L); hemoglobin, 13.3 g/dL (reference, 13.0 –17.4 g/dL); platelets, 61 X $10^9$/L (reference, 140 X $10^9$/L to 440 X $10^9$/L)]. The physical examination was unremarkable, as were routine laboratory tests. A bone marrow (BM)[6] aspirate and biopsy demonstrated hypocellularity (20%) without dysplasia. Results for BM cytogenetics, fluorescence in situ hybridization (FISH), and a diexpoybutane test for Fanconi anemia were normal. Flow cytometry findings for peripheral blood showed no lymphoproliferative or myeloid-maturation disorder, nor any protein defect consistent with paroxysmal nocturnal hemoglobinuria. Over the next 5 years, the patient remained asymptomatic and transfusion free with stable blood counts and an unchanged BM histology.

As part of a review of potential therapeutic interventions, he sought a second opinion, at which time the patient recounted a history of recurrent urethral strictures from ages 6 to 9 years and again at age 22 years. His hair had started to gray at 11 years, and his hairline to recede by age 16. There was a family history of similar findings. He described his fingernails and toenails as always "embarrassingly" dry and cracked. He recounted having had excess tearing and that friends would note he was crying although he was unaware of tear production. On examination, the patient had hair thinning, graying, and mild frontal balding. He had subtle, nonblanching, slightly reddish brown reticular pigmentation over his upper anterior and posterior thorax. All of his nails were markedly dystrophic. There was a flat 1 X 1.5 cm white lesion on the upper hard palate. The remainder of his examination was unremarkable. His blood counts had been quite stable over the 5 antecedent years: white blood cell count, 1.4 X $10^9$/L to 3.8 X $10^9$/L (reference, 4.2 X $10^9$/L to 9.9 X $10^9$/L); absolute neutrophil count, 0.71 X $10^9$/L to 2.7 X $10^9$/L (reference, 2.4 X $10^9$/L to 7.6 X $10^9$/L); hemoglobin, 12.9 –14.5 g/dL (reference, 13.0–17.4 g/dL), with marked macrocytosis (mean corpuscular volume, 105–111 fL; reference, 82.0–100 fL); and platelets, 46 X $10^9$/L to 71 X $10^9$/L (reference, 140 X $10^9$/L to 440 X $10^9$/L).

| Questions to Consider |
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| • What are the causes of aplastic anemia (AA)[6]? |
| • Given this patient's symptoms, what might be the diagnosis? |
| • What laboratory test would be useful for the diagnosis? |

[6]Nonstandard abbreviations: BM, bone marrow; FISH, fluorescence in situ hybridization; AA, aplastic anemia.

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**Final Publication and Comments**

The final published version with discussion and comments from the experts will appear in the January 2013 issue of *Clinical Chemistry*. To view the case and comments online, go to http://www.clinchem.org/content/vol59/issue1 and follow the link to the Clinical Case Study and Commentaries.

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