Clinical Case Study

Increased Homocysteine in a Patient Diagnosed with Marfan Syndrome

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CASE

A 53-year-old Caucasian woman was diagnosed in late childhood with Marfan syndrome according to characteristic skeletal features and bilateral lens dislocation. In addition, she has a history of nonischemic cardiomyopathy with severe left ventricular failure and atrial fibrillation, diabetes mellitus type 2, hyperlipidemia, progressive dementia, numbness in the lower extremities, and hypothyroidism following thyroidectomy for thyroid cancer. Additional findings revealed in a physical examination included an upper-to-lower segment ratio of 0.88 (an upper-to-lower segment ratio < 0.85 and arm span-to-height ratio > 1.05 are two of the diagnostic criteria for Marfan syndrome), an arm span–to–height ratio of 1.02, an elongated face, a high arched palate, and crowded dentition. She recently underwent further laboratory testing after a cardiologist did not find 2 characteristic features of Marfan syndrome, namely an enlarged aortic root and mitral valve prolapse. Her total plasma homocysteine and methionine concentrations were increased at 198 μmol/L (reference interval, 5–15 μmol/L) and 370 μmol/L (reference interval, 10–50 μmol/L), respectively. The patient's plasma homocystine concentration was 48 μmol/L (reference interval, <2 μmol/L), and her urine homocystine concentration was also markedly increased. These biochemical abnormalities are not characteristic of Marfan syndrome. Her diagnosis was reconsidered in light of these new data.

Questions to Consider
• What are the molecular defects responsible for Marfan syndrome?
• What pathologic conditions are associated with lens dislocation?
• What conditions are associated with increased homocysteine in blood and urine?

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the November 2010 issue of *Clinical Chemistry*. To view the case and comments online, go to http://www.clinchem.org/content/vol56/issue10 and follow the link to the Clinical Case Study and Commentaries.

Educational Centers

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