

## Increased Homocysteine in a Patient Diagnosed with Marfan Syndrome

Olajumoke Oladipo,<sup>1</sup> Laurie Spreitsma,<sup>2</sup> Dennis J. Dietzen,<sup>1,2\*</sup> and Marwan Shinawi<sup>2</sup>

<sup>1</sup> Department of Pathology and Immunology and <sup>2</sup> Department of Pediatrics, Washington University School of Medicine, St. Louis, MO.

\* Address correspondence to this author at: Department of Pediatrics, Box 8116, Washington University School of Medicine, One Children's Place, St. Louis, MO 63110. Fax 314-454-2274; e-mail Dietzen\_d@kids.wustl.edu.

### 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  $\mu\text{mol/L}$  (reference interval, 5–15  $\mu\text{mol/L}$ ) and 370  $\mu\text{mol/L}$  (reference interval, 10–50  $\mu\text{mol/L}$ ), respectively. The patient's plasma homocysteine concentration was 48  $\mu\text{mol/L}$  (reference interval, <2  $\mu\text{mol/L}$ ), and her urine homocysteine 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

If you are associated with an educational center and would like to receive the cases and questions 1 month in advance of publication, please email [clinchem@aacc.org](mailto:clinchem@aacc.org).

AACC is pleased to allow free reproduction and distribution of this Clinical Case Study for personal or classroom discussion use. When photocopying, please make sure the DOI and copyright notice appear on each copy.

All previous Clinical Cases Studies can be accessed and downloaded online at <http://www.aacc.org/resourcecenters/casestudies/>.

---

AACC is a leading professional society dedicated to improving healthcare through laboratory medicine. Its nearly 10,000 members are clinical laboratory professionals, physicians, research scientists, and others involved in developing tests and directing laboratory operations. AACC brings this community together with programs that advance knowledge, expertise, and innovation. AACC is best known for the respected scientific journal, *Clinical Chemistry*, the award-winning patient-centered web site *Lab Tests Online*, and the world's largest conference on laboratory medicine and technology. Through these and other programs, AACC advances laboratory medicine and the quality of patient care.