An Unusual Electrophoresis

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CASE DESCRIPTION

A 41-year-old white male patient presented to the pulmonary clinic complaining of chronic exertional dyspnea. He had a 15-year history of smoking, but no prior diagnosis of lung disease. A comprehensive battery of laboratory tests was performed. Results for complete blood count, serum chemistry panel, hepatic function tests, and inflam-

References

matory markers were within reference intervals. Arterial blood gas measurement revealed hypoxemia and hyper-capnia. Serum protein capillary zone electrophoresis results are shown in Fig. 1.

QUESTIONS
1. What finding does this protein electrophoresis gel demonstrate?
2. What could be the diagnosis for this pattern?
3. How can the diagnosis be established?

The answers are below.

ANSWERS

The patient’s serum α-1 globulins were reduced at 1.3 g/L (reference interval, 2.1–3.5 g/L). Potential causes include malnutrition, excessive excretion, decreased production, and α1-antitrypsin (A1AT) deficiency. The clinical history was consistent with A1AT deficiency (1–3). The diagnosis should be confirmed by direct quantification of circulating A1AT by immun assay and confirmation of the presence of A1AT disease alleles by isoelectrofocusing and/or genotyping (2–4). In this case, A1AT was not detectable and a Z/Z homozygote phenotype was found.

Author Contributions: All authors confirmed they have contributed to the intellectual content of this paper and have met the following 3 requirements: (a) significant contributions to the conception and design, acquisition of data, or analysis and interpretation of data; (b) drafting or revising the article for intellectual content; and (c) final approval of the published article.

Authors’ Disclosures or Potential Conflicts of Interest: No authors declared any potential conflicts of interest.

Acknowledgments: We thank I. Cuvelier, CERBA Laboratory.

References

News & Views

Realities of Observational Study Registration

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Interest in registration of observational studies is growing. Observational studies (OSs)³ of human subjects represent a large fraction of published scientific reports and may be considered closer to bedside than clinical trials (CTs), yet the majority are unregistered in prospective study registries. A recent article by Dal-Re et al. in Science Translational Medicine (1) provides commentary on the ethical and scientific rationale supporting prospective registration of OSs. To briefly illustrate their point, the authors performed a literature search of PubMed articles from 2011 and observed that the number of publications from OSs outnumbered those from randomized controlled trials (RCTs) almost 300,000 to 20,000. Although approximately 20% of RCT reports were accompanied by a registration num-

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DOI: 10.1373/clinchem.2014.226258
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3 Nonstandard abbreviations: OS, observational study; CT, clinical trial; RCT, randomized controlled trial.