



An Unusual Cause of Physical Disability and Vascular Complications in a Young Man

Cekerevac I^{1,2}, Susa R², Stevanovic D^{1,3} and Simovic S^{1,3*}

¹Department of Internal Medicine, University of Kragujevac, Serbia

²Clinic for Pulmonology, Clinical Center Kragujevac, Serbia

³Clinic for Cardiology, Clinical Center Kragujevac, Serbia

Abstract

A 17-year-old male with fatigue, diagnostic tests revealed multiple segmental embolization. Treatment with oral anticoagulants was started. Two years later, patient was re-admitted with sub-massive pulmonary embolism, genetic testing revealed AKT1 mutation (Proteus syndrome). Three years later, patient was admitted again and massive pulmonary embolism was established as a cause of death.

Case Study

A 17-years-old male presented with fatigue. Medical history revealed port-wine stains (Figure 1A) and an amputation of the left leg when he was 3 years old. Examination revealed kyphoscoliosis (Figure 1B), hyperostosis of the temporal bone (Figure 1C), increased Systolic Pressure of Right Ventricle (RVSP) (45 mmHg) (Figure 1D). CTPA revealed bullous formation (Figure 1E) and segmental embolization's (Figure 1F). Treatment with oral anticoagulants with was started. Two

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*Correspondence:

Stefan Simovic, Clinic for Cardiology,
Clinical Center Kragujevac, Zmaj Jovina
30, 34000 Kragujevac, Serbia, Tel:
+381644111120;

E-mail: simovicst@gmail.com

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Figure 1: 1A) Port wine stains on the abdomen of the patient; 1B) Kyphoscoliosis of the thoracic spine; 1C) hyperostosis of the pyramid extension of the temporal bone; 1D) Echocardiographic measurement of systolic right ventricular pressure of 45 mmHg; 1E) Large-scaled bullous formation seen on CT; 1F) Multiple, segmental pulmonary embolization; 1G) Secondary sub-massive pulmonary embolism with left-sided pleural effusion; 1H) Cerebriform lesions on the left hand with clinodactyly; 1I) Cerebriform lesions on the right foot.

years later, he presented with chest pain and dyspnea. CTPA showed sub-massive pulmonary embolism (Figure 1G). He had cerebriiform lesions on the left hand (with clinodactyly) and right foot (Figure 1H, 1I). Genetic testing was done revealing AKT1 mutation indicating Proteus syndrome [1]. Three years later presented with hypotension and respiratory failure, RVSP was increased (70 mmHg), so patient received thrombolytics. The patient died before CTPA was performed, but given the course, massive pulmonary embolism was established as a cause of death.

This is first-of-its-kind report of a patient with Proteus syndrome and recidivant pulmonary embolism, despite intensified anticoagulation.

Teaching Points

- Proteus syndrome can cause recidivant and fatal pulmonary embolism, despite intensified anticoagulation.

References

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