

Unrecognized type II respiratory failure with TTN gene mutation in elderly

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Introduction: Patients with type II respiratory failure are unable of elimination of CO₂ with venting failure. Causes of type II respiratory failure could be various reasons, including pulmonary problem, thoracic wall abnormality, central nervous system or neuromuscular disease. In elderly, the most common cause of type II respiratory failure usually related to chronic pulmonary problems. However, hereditary respiratory failure in elderly is not common.

Case: 69-year-old man admitted to the hospital for operation of vitrectomy. He had a past medical history of hypertension, diabetic mellitus since 1 year ago. His body mass index was 32.5. On arrival at ward, his temperature was 36.8°C; heart rate, 88 beats/minute; blood pressure, 140/70 mmHg; respiratory rate, 20 breaths/minute; and he had an oxygen saturation of 85%. Without any respiratory symptom, blood gas analysis was performed. Arterial pH was 7.28, Pa CO₂ was 74.6 mmHg (9945.85Pa) and PaO₂ was 52.2 mmHg (6959.429Pa). After noninvasive ventilator applied by critical care team, arterial Pa CO₂ was reached to 48.3 mmHg with 7.35 of pH. He had never smoked neither history of respiratory disease. To figure out the respiratory failure, he underwent tests for respiratory problems. Pulmonary function test (PFT) showed severe restrictive pattern (forced vital capacity, 0.96 liter, predicted 25%) and severe decreased diffusion capacity (37%). Elevated of both diaphragms with passive atelectasis was revealed in chest computed tomography (CT). On his familial history, we came to know that patient's younger sister had history of gene mutation disease (A), (Figure 1) revealed as TTN Gene likely pathogenic variant. Finally, he was confirmed with TTN Gene mutation as well (figure 2).

Conclusion: TTN gene related respiratory failure is known as hereditary myopathy with early respiratory failure (HMERF). HMERF is characterized by early respiratory insufficiency with limb muscle weakness. HMERF is usually diagnosed before in patient's 50s. However unrecognized respiratory failure with or without neurologic symptoms could be the first sign of TTN gene mutation even though in elderly.

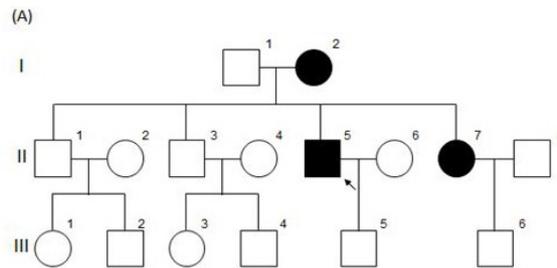


Figure 1. Family tree of patient.

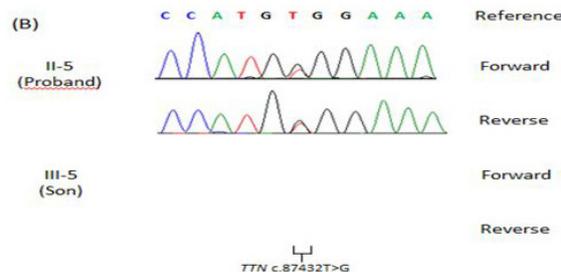


Figure 2. TTN Gene variation c.87432T>G.