Two novel mutations in the gene that codes for acid α-glucosidase in a baby with Pompe disease

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An 8-month-old baby was admitted to our hospital because of prolonged fever and severe pneumonia. Clinical examination displayed mild hepatomegaly, protruding tongue and hypotonia (Panel A). Blood analysis showed increased ALT 144 U/L (n = 0–40), AST 270 U/L (n = 5–34) and CK 696 U/L (n = 29–200). Chest X-ray revealed a large heart. Electrocardiography showed a short PR interval and severe left ventricular hypertrophy with ST-T changes. Echocardiography confirmed a severe form of biventricular hypertrophy, left ventricular papillary muscles were severe hypertrophy so that left ventricular cavity was out of shape and nearly obliterated. M-mode image at papillary muscle level showed severe hypertrophy of the septum and left ventricular posterior wall (Panels B – E). The assay for acid α-glucosidase (GAA) from whole blood using dried blood spot filter paper was done. The activity of GAA was 2.0 (normal range: 62.3–301.7 nmol/h/mgPr), so he was diagnosed with Pompe disease. Pompe disease is transmitted as an autosomal recessive trait and could be the cause of hypertrophy cardiomyopathy. It is caused by mutations in the gene encoding the GAA, located on chromosome 17q25.2–q25.3. A molecular genetic analysis revealed two novel compound heterozygous missense mutations in GAA: c.1385T>C (p.Leu462Pro) and c.1669A>T (Ile557Phe) (Panel F, arrows). The two mutations detected in this baby have never been recorded in the human genome database before, so they represent new mutations in the GAA. Unfortunately, 2 months later, the baby was death because of heart failure and acute severe bronchial pneumonia.

Supplementary material is available at European Heart Journal online.

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