We report the case of a full term male child with multiple organ dysfunctions caused by mutations of the TTN gene. Our patient had cardiac abnormalities verified by fetal echocardiography at 26 weeks of gestational age, when fetal hydrops due to cardiac decompensation was shown.

Due to specific appearance seen at birth — generalized oedema, light facial and sexual dimorphism with hypogenitalism and cryptorchidism, axial hypotonia in addition to the proven congenital heart defects, a genetic disorder or syndrome with muscle involvement was suspected. Multiple comorbidities were present since birth, mainly cardiac and neurologic, some of which have been described in patients with core myopathies (atrial and ventricular septal defects, supraventricular tachycardia, hypotonia, developmental delay, myopathy). Due to the complexity of the heart defect at the age of 2 months a partial pulmonary trunk banding and ligation of the ductus Botalli (PDA) was initially carried out, and at 9 months a complete correction of heart defect was performed. Our patient also had failure to thrive as well as recurrent vomiting, which was later on associated with extraluminal compression of the proximal duodenum, intestinal malrotation and malfixation. Although a surgical approach somewhat increased tolerance of peroral food intake, chronic diarrhoea and failure to thrive persisted.

In addition, our patient had several metabolic and endocrine disorders including hypocalcaemia, hypomagnesaemia, hyponatremia, repeatedly elevated levels of PTH, hypothyroidism etc. Our patient had recurrent, mainly respiratory infections with multisresistant microorganisms, and required intubation and long-lasting mechanical ventilation. The above mentioned multiple disorders of various organ systems, with complications caused by multiple sepses, ultimately resulted in multiorgan failure, and despite all the applied treatment at the ICU, resulted in a lethal outcome. Corresponding findings described in other patients with core myopathies included neonatal hypotonia, poor sucking, severe motor skill delay, complex heart defects and cardiomyopathy.

A five year-old girl came to the tourist ambulance because of the dysuria, sore throat and tingling of external genitalia. A local physician prescribed amoxicillin/clavulanic acid suspension. She took her first dose in the afternoon. During the night, parents noticed the rash and redness of the entire body. During the night, another dose of antibiotic was given, after which the rash and redness continued to spread rapidly.

Immediately methylprednisolone and chloropromazine are administered intramuscularly. Upon arrival to the hospital, the girl was a normal state of consciousness with dyspnea, breathing frequency 25/min, SpO2 97%, dehydrated, febrile 38.1 °C, tachycardic (135/min) and normotensive (RR 103/66 mmHg). On the skin of the face and on the larger surface of the body (TBSA = 80%) were visible bullae and vesicles which ruptured and it came to skin peeling. Due to the progression of respiratory insufficiency, tracheotomy was performed. Also due to the increase in inflammatory parameters for the first ten days, ceftriaxone and amikacin were ordered, with all supportive therapy. On the second day of admission, cyclosporine is ordered. All the swabs were negative. Lyell’s syndrome, or toxic epidermal necrolysis, is a rare, potentially life-threatening mucocutaneous disease, usually provoked by the administration of a drug and characterized by acute necrosis of the epidermis. The drugs most frequently incriminated are nonsteroidal antiinflammatory drugs, chemotherapeutics, antibiotics, and anticonvulsants. Although the cases where amoxicillin/clavulanic acid suspension caused this condition were described, in our case, amoxicillin/clavulanic acid suspension was probably not the cause, given that the girl had been treated twice in the past with this drug without side effects.