Background Cytochrome P450 Oxidoreductase (POR) deficiency is one of the rare forms of adrenal insufficiency. It causes suspicious genitalia in boys. The disorder in steroid synthesis carries the symptoms of both 21-hydroxylase and 17–20-lyase deficiency. Cortisol deficiency is not frequent, but the stress response is impaired.

Case An 8-month-old male patient was admitted to our hospital with vomiting for 15 days. Hyponatremia and hyperkalemia were detected in the first laboratory tests of the patient and he was hospitalized for further investigation. In the background: The patient was born at term by caesarean section. His birth weight was 3600 grams, developmental stages were normal and he had no hospitalization before. Family history: the parents were healthy and they were unrelated. Physical examination; Height: 65 cm (-2.23 SDS; <3p), weight: 6.6 kg (-3.01 SDS; <3p), head circumference: 43 cm (-1.53 SDS; 3–10p). Blood pressure: 100/62 mmHg, pulse: 130/min, respiration rate was 35/min. The general condition was moderate, the skin turgor was decreased and the skin was pale. His systemic examinations were normal, externally in male view but he had hypospadias and bilateral descended testes. The laboratory tests are given in table 1. Abdominal ultrasound was normal. Scrotal ultrasound showed bilateral testes in inguinal canal. Adrenal insufficiency was considered in the preliminary diagnosis. Performed ACTH stimulation test’s results are in table 2.

Results The patient is considered to have POR deficiency and his genetic research is in progress. Hydrocortisone 15 mg/m²/day and fludrocortisone 0.1 mg/day were prescribed. We presented this case for discussing POR deficiency as a cause of rare adrenal insufficiency.