Imerslund-Grasbeck syndrome is an autosomal recessive disorder characterized by proteinuria and megaloblastic anemia due to selective deficiency in absorption of vitamin B12 (cobalamin). As vitamin B12 is stored in fetus by maternal-fetal transport, symptoms and signs of deficiency are not apparent just after birth. Two cases of Imerslund-Grasbeck syndrome were presented because of its rarity.

Case I: A 21 months old girl presented with symptoms of fever, weakness and pallor. Her parents were first degree relatives. In physical examination; weight was <3p and height was 25p. She had 2cm of hepatomegaly. Neuromotor development and other physical findings were normal. In laboratory findings; complete blood count revealed anemia (Hb:5.8g/dl, Hct:17.6%, MCV:115fl). Blood smear and bone marrow aspiration biopsy showed megaloblastic anemia. Levels of serum vit B12 and folate were 60pg/ml (200-950) and 19ng/ml (4.1-20.4), respectively. 24-hour urine protein excretion was 19.3mg/m2/hr.

Case II: A 26 months old girl presented with symptoms of pallor, cough, fever and weakness. Her parents were 2nd degree relatives. In physical examination both weight and height percentiles were 3p. She had mild periorbital edema with blood pressure in normal range. Neuromotor development and other physical findings were normal. Complete blood count revealed anemia (Hb:3.8g/dl, Hct:12%, MCV: 127fl). Blood smear and bone marrow aspiration biopsy showed megaloblastic anemia. Levels of serum vit B12 and folate were 38pg/ml (200-950) and 15ng/ml (4.1-20.4), respectively. 24-hour urine protein excretion was 220mg/m2/hr.

Both of the patients were investigated for probable renal pathology but no abnormality was detected. In this way, they were diagnosed as Imerslund-Grasbeck syndrome in view of clinical and laboratory findings.

In conclusion; concurrence of megaloblastic anemia and proteinuria in societies where consanguinous marriage is widespread, should suggest this syndrome.