SUPPLEMENTAL DATA

Detailed investigations of proximal tubular function in Imerslund-Gräsbeck Syndrome

CLINICAL DATA ON INVESTIGATED PATIENTS

Abbreviations: MCV (mean corpuscular volume); $P_{\text{vitB12}}$ (plasma vitamin $B_{12}$)

Family 1 (patient A):
Is a 6-year-old male, diagnosed at 18 months with anaemia and diarrhoea,
$P_{\text{vitB12}}$: 49 pg/l, folic acid: normal
Anaemia: MCV 97 µm$^3$
Kidney: proteinuria of 2.16 g/l including microalbuminuria of 1040 mg/l, no homocysteine or amino acids was detected in the urine.
Treatment: vitamin $B_{12}$ injections

Family 2 (patient A):
Is a 16-year-old female
$P_{\text{vitB12}}$: < 100 pg/l, folic acid: normal
Anaemia: MCV 87 µm$^3$
Kidney: proteinuria of 1.23 g/l including microalbuminuria of 0.934 mg/l, no homocysteine or amino acids was detected in the urine.
Treatment: vitamin $B_{12}$ injections

Family 2 (patient B):
Is a 27-year-old male.

Family 3 (patient A):
Is a 6-year-old female diagnosed with anaemia.
$P_{\text{vitB12}}$ < 100 pg/l, folic acid: normal
Kidney: proteinuria 1.45g/l, microalbuminuria; 900 mg/l, no homocysteine or amino acids was detected in the urine.
Treatment: vitamin $B_{12}$ injections
Family 4 (patient A and B):

Two 6-year-old, female, non-identical twins diagnosed when they were 2 years old with megaloblastic anaemia and low-molecular-weight proteinuria.

\[ P_{\text{vitB12}}: < 50 \text{ pg/ml for both patients.} \]

Treatment: They are treated weekly per os with 10 µg cyanocobalamin and are currently asymptomatic with normal growth. Kidney function was normal at age 5 years (serum creatinine: 0.41 mg/dl and 0.36 mg/dl; eGFR (ml/min/1.73m\(^2\)): 141 and 159) but low-molecular weight proteinuria persists.

Family 5 (patient A):

Is a 10-year-old male diagnosed at age 3 years with anaemia and diarrhoea.

\[ P_{\text{vitB12}}: < 60 \text{ pg/l, folate: normal} \]

Anaemia: MCV 108 µm\(^3\)

Kidney: proteinuria 3.14 g/l and low homocysteine in the urine but normal serum creatinine of 0.3 mg/dl.

Treatment: vitamin B\(_{12}\) injections and vitamin D supplementation.

Family 5 (patient B):

Is a 6-year-old male diagnosed at age 18 months with anaemia, urinary infections, and kyphosis of the lumbar region.

\[ P_{\text{vitB12}}: < 60 \text{ pg/l, folic acid: normal} \]

Anaemia: MCV 80 µm\(^3\)

Kidney: low proteinuria and normal serum creatinine of 0.3 mg/dl.

Treatment: vitamin B\(_{12}\) injections and vitamin D supplementation.

Family 6 (patient A):

Is a 4-year-old female diagnosed with retarded growth at the age of 3 years with low \( P_{\text{vitB12}} \) as well as low plasma folic acid.

\[ P_{\text{vitB12}}: 21 \text{ pg/ml} \]

Folic acid: 60.9 ng/ml

Anaemia: MCV 105 µm\(^3\), haemoglobin 5.4 g/dl.

Kidney: no proteinuria

Treatment: vitamin B\(_{12}\) injections every 3 months Cohemin\(^\circledast\). Initially, folic acid supplements was also administered but is no longer needed.