

| Patient ID | 1.1 | 1.2 | 1.3 | 1.4 | 2.1 | 2.2 | 3.1 | 4.1 | 5.1 |
|-----------------------------------|----------------------|-------------------------|-------------------------|-------------------------|--------------------|--------------------|--------------------|--------------------|--------------------|
| Sex | Male | Female | Female | Female | Female | Male | Male | Male | Female |
| Mutation | | | | | | | | | |
| Nucleotide | c.-167G>T/c.-167G>T | c.-167G>T/c.-167G>T | c.-167G>T/c.-167G>T | c.-167G>T/c.-167G>T | c.-167G>T/c.422G>A | c.-167G>T/c.422G>A | c.-167G>T/c.470T>C | c.-167G>T/c.422G>A | c.-167G>T/c.422G>A |
| Predicted protein | p.? | p.? | p.? | p.? | p.?/p.Arg141His | p.?/p.Arg141His | p.?/Phe157Ser | p.?/p.Arg141His | p.?/p.Arg141His |
| Hyperinsulinism | | | | | | | | | |
| Birth weight [g] | 2880 | 3400 | 3000 | 3750 | 3850 | 3400 | 3325 | 2960 | N/A |
| Age at diagnosis [months] | 6 | 10 | 48 | 10 | 0 | 8 | 21 | 0 | 4 |
| Treatment | none | none | none | none | Diazoxide | Diazoxide | Diazoxide | Diazoxide | N/A |
| Renal disease | | | | | | | | | |
| Antenatal abnormalities | cysts/polyhydramnios | no | no | no | no | no | no | no | N/A |
| Age at renal diagnosis [months] | 0 | 6 | 48 | 11 | 48 | 0 | 21 | 0 | 36 |
| eGFR at diagnosis [ml/min/1.73m2] | 70 | 100 | 85 | 115 | 95 | 90 | 90 | 50 | 95 |
| Kidney size [percentile] | >95th | >95th | >95th | >95th | >95th | >95th | >95th | >95th | >95th |
| Morphology | cysts | hyperechogenicity/cysts | hyperechogenicity/cysts | hyperechogenicity/cysts | cysts | cysts | cysts | cysts | cysts |
| Age at last f/u [years] | 10 | 15 | 20 | 14 | 11 | 4 | 1 | 2 | 14 |
| eGFR [ml/min/1.73m2] at last f/u | 70 | 90 | <10 (18) | 75 | 80 | 40 | 90 | 50 | 85 |
| Liver disease | | | | | | | | | |
| Morphology (ultrasound) | normal | normal | normal | normal | cysts | cysts | normal | normal | coarse echotexture |
| Liver function tests | normal | normal | normal | normal | normal | normal | normal | normal | N/A |

| Patient ID | 6.1 | 7.1 | 8.1 | 9.1 | 9.2 | 10.1 | 10.2 | 11.1 |
|-----------------------------------|--------------------|----------------------|----------------------|--------------------|--------------------|--------------------|--------------------|--------------------|
| Sex | Male | Female | Female | Male | Male | Female | Male | Female |
| Mutation | | | | | | | | |
| Nucleotide | c.-167G>T/c.422G>A | c.-167G>T/c.422G>A | c.-167G>T/c.255+1G>A | c.-167G>T/c.620T>C | c.-167G>T/c.620T>C | c.-167G>T/c.470T>C | c.-167G>T/c.470T>C | c.-167G>T/c.470T>C |
| Predicted protein | p.?/p.Arg141His | p.?/p.Arg141His | p.? | p.?/p.Phe207Ser | p.?/p.Phe207Ser | p.?/p.Phe157Ser | p.?/p.Phe157Ser | p.?/p.Phe157Ser |
| Hyperinsulinism | | | | | | | | |
| Birth weight [g] | 4200 | 4252 | 4500 | 3440 | 3450 | N/A | 3720 | N/A |
| Age at diagnosis [months] | 0 | N/A | 12 | 14 | 0 | 18 | 11 | 14 |
| Treatment | Diazoxide | Diazoxide/cornstarch | N/A | Diazoxide | Diazoxide | N/A | N/A | Diazoxide |
| Renal disease | | | | | | | | |
| Antenatal abnormalities | cysts | N/A | N/A | cysts | cysts | N/A | N/A | no |
| Age at renal diagnosis [months] | 0 | 36 | 15 | 0 | 0 | N/A | N/A | 20 |
| eGFR at diagnosis [ml/min/1.73m2] | 20 | 120 | 75 | 30 | 40 | N/A | N/A | 105 |
| Kidney size [percentile] | >95th | >95th | >95th | N/A | N/A | N/A | N/A | >95th |
| Morphology | cysts | cysts | cysts | cysts | cysts | cysts | cysts | cysts |
| Age at last f/u [years] | 6 | 11 | 7 | 4 | 0.3 | N/A | N/A | 11 |
| eGFR [ml/min/1.73m2] at last f/u | <10 (2) | 141 | 40 | 65 | 70 | N/A | N/A | 94 |
| Liver disease | | | | | | | | |
| Morphology (ultrasound) | cysts | cysts | normal* | normal | normal | cysts | N/A | cysts |
| Liver function tests | normal | normal | normal | normal | normal | normal | normal | N/A |

Table 1: Clinical Details

Shown are pertinent clinical data for the 17 patients. Note the presence of the PMM2 promoter mutation c.-167G>T in all patients.

N/A: not available; f/u: follow-up; eGFR: estimated glomerular filtration rate. * Ultrasound imaging of the liver was normal, yet a biopsy revealed ductal plate malformation (see Figure 3).