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# P440 -Treatment of Glycogen Storage Disease Type 1a by combined Liver and Kidney Transplantation

Doctor Mukesh Kumar[1], Doctor Laurie Solomon[1], Professor Christian Hendriksz[2], Doctor Karolina Stepien[2], Professor Peter Lodge[4], Professor Lee Claridge[3], Advanced Clinical Practitioner Diane Green[2], Doctor Audrey Dillon[3]

*[1]Regional Renal Centre, Royal Preston Hospital, Preston, United Kingdom, [2]Adult Inherited Metabolic Diseases Department, Salford, United Kingdom, [3]Liver Unit, St James's University Hospital, Leeds, United Kingdom, [4]HPB and Tranplant Unit, St James's University Hospital, Leeds, United Kingdom*

Introduction: Glycogen storage disease type 1a (GSD 1a) is an autosomal recessive condition characterized by deficiency of glucose-6-phosphatase resulting in excessive deposition of glycogen and lipid in the liver and kidney. It results in spontaneous hypoglycaemia and requires treatment with regular carbohydrate meals and overnight administration of feed, rich in protein, fibre and fructo-oligosaccharides. Renal involvement with proteinuria and progressive kidney failure may develop in childhood, but there are very few reports in young adults. Angiotensin converting enzyme inhibitors or angiotensin II receptor blockers are recommended for proteinuria. Liver and kidney transplantation has rarely been described, but is the only treatment with potential to correct biochemistry and limit lifelong complications.

Case report: A 6-month old Asian girl presented with failure to thrive. She was diagnosed with GSD 1a (exon 1-3 deletion) when under the care of the pediatric metabolic unit. At 17 she transitioned to adult renal services, when she had renal impairment and heavy proteinuria (urea 16.4 mmol/l, creatinine 112 µmol/l, urinary protein: creatinine ratio 491 mg/mmol). She had been treated since the age of three with frequent meals containing cornstarch during the day, overnight PEG feeds and enalapril for proteinuria. Metabolic control was poor. Investigations included, cholesterol 12.4 mmol/l, triglycerides 35 mmol/l and serum bicarbonate was 15.5 mmol/l. Urine pH >5.5 and urinary citrate <50 µmol/l indicated distal renal tubular acidosis. Ultrasound scan showed a normal appearance of both kidneys. Renal biopsy showed 3/7 sclerosed glomeruli, moderate tubular atrophy and interstitial fibrosis. Periodic Acid-Schiff with diastase stain confirmed glycogen accumulation in proximal tubules. Despite treatment with enalapril, atorvastatin, sodium bicarbonate and allopurinol, kidney function deteriorated. She had haemodialysis for four months at age 21 before receiving a liver and kidney transplant from a deceased donor. Immunosuppression comprised tacrolimus and mycophenolate mofetil.

Result: Her post-transplant course was uneventful with immediate function of liver and kidney. Overnight feeding was discontinued. Her PEG tube was removed for the first time after 20 years. One year after transplantation results include haemoglobin 112 g/l, urea 5.9 mmol/l, bicarbonate 21.7 mmol/l, creatinine 121 µmol/l, eGFR 55 ml/min, cholesterol 2.6 mmol/l, triglycerides 1.15 mmol/l, ALT 26 IU/L, bilirubin 13 µmol/l and alkaline phosphatase 81 IU/L.

Discussion: The optimum management of end-stage kidney failure in GSD1a is unknown. Combined kidney and liver transplantation is a major undertaking but provides the best chance of biochemical correction. Our patient had an excellent outcome with resolution of hypoglycaemia, improved metabolic control and quality of life.