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Renal transplant in Carbohydrate-deficient glycoprotein syndrome type 1a.

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Introduction:

Carbohydrate-deficient glycoprotein syndrome (CDG) is a group of rare metabolic disorders which affect glycosylation, with over 80 subtypes.[1,2] The disease typically presents from birth or infancy involving multiple organs with a wide range of signs and phenotypes. This can range from hepatosplenomegaly, hepatic impairment, cardiomyopathy, pericardial effusions and renal impairment as well as bleeding and clotting abnormalities. [1,2] There is limited evidence in the literature of transplantation in this patient population, with only single case reports of heart and liver transplantation (3,4).

Case description:

We present the case of a 14year old boy with CDG type 1a who underwent a successful renal transplant. He was diagnosed with CDG type 1a within the first 6 months of life. His phenotype includes global development delay, renal failure secondary to diffuse mesangial sclerosis, visual impairment and hepatomegaly. In addition to the recognised clotting abnormalities of CDG of an unpredictable risk of thrombosis or bleeding, on an extended clotting screening our patient also had a low protein C and anti-thrombin 3, and a low factor XI.

The proteinuria from his renal disease led to progressive renal impairment cumulating in the need for renal replacement therapy as peritoneal dialysis at 12 years of age and listing for transplantation. However, he had an extensive bleed following the insertion of a peritoneal catheter and was subsequently suspended from the transplant list pending further exploration of his bleeding risk.

Historical surgical procedures (liver biopsy and gastrostomy insertion) had not involved significant bleeding. Local haematological and secondary centre surgical/haematological opinions were sought in conjunction with the family's wishes to evaluate and attempt to quantify the excess risks involved in renal transplantation. In particular the risk of graft thrombosis and death during the procedure from bleeding. The risk was unquantifiable and through shared decision making with the family the patient was listed for transplantation.

To minimise the risk of bleeding fresh frozen plasma (FFP) was given 1 hour before transplantation. We had factor XI on site and if there was unexpected bleeding an urgent level would be taken and if <45iug/dL to give 5iu/kg infusion with an urgent re-check as a level >70iu/dL increases the risk of thrombus. The recommended anti-coagulation plan post theatre was for IV heparin once "risk of bleeding decreased" as opposed to enoxaparin. To minimise the risk of thrombosis perioperatively good hydration was maintained and it was advised to have "expert surgery" to reduce risk of vessel thrombosis. The transplant was uncomplicated with successful primary graft function. The patient had a complicated post-operative recovering including a significant gastrointestinal bleed.

Discussion:

We believe this is the first renal transplantation in a patient with CDG. Shared decision making is essential with families and patients with CDG due to the rare nature and limited understanding of the disease as risks will be unpredictable and unquantifiable. A collaborative approach across centres and specialities was instrumental in allowing a balanced management plan for the risk of bleeding and thrombosis to be expediently managed.