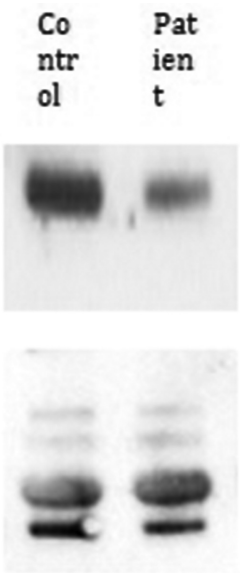


**Abstract G477 Figure 2** PAR2 expression in serum from control and patient samples


**Abstract G477 Figure 3** PAR2 expression in immortalised kidney cells,demonstrating expression in wild-type endothelial,mesangial and podocyte cells

whether PAR2 represents a unique pathway in these rare renal phenotypes.

## G478(P) A CASE REPORT OF TRBETA MUTATION LEADING TO RAISED T4 LEVELS

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**Introduction** We present a five year old girl with thyroid hormone resistance, subsequently discovered to be heterozygous for TRbeta mutation. This case highlights the necessity to investigate, in detail, all children with persistently high thyroxin with normal TSH levels in order to aid future management and the necessity to follow them up.

**Case report** The patient was born at term by normal delivery, weighing 3.34kg (50th centile). She was referred at six months of age for poor weight gain (2nd centile). Blood tests showed an elevated free T4 (43.4) with normal TSH (3.10). Systemic examination was normal and remained so over the following months. Repeat thyroid function tests showed persistently elevated T4 with normal TSH. At 2 ½ years old, genetic analysis revealed she is heterozygous for TRbeta mutation (thyroid hormone receptive gene). Mum has no mutation detected and her father cannot be tested for unavoidable reason. Since then, she has had slow growth, idiopathic thrombocytopenic purpura, vitamin D deficiency, and coeliac disease. Broader antibody testing has not revealed an underlying autoimmune aetiology to date. Recently, the patient has been investigated for recurrent falls and abnormal gait. She has right sided hemi hypotrophy with drooped shoulder and pelvis, along with winged scapulae, flared ribs and prominent abdomen. Her gross motor skills are generally delayed.

**Conclusion** Mutation of the beta thyroid hormone receptor is usually either autosomal dominantly inherited or is a *de novo* mutation, resulting in defective patterns of gene expression. This is a rare disorder, usually presenting with goitre. TRbeta mutation should be considered in children with persistently elevated T4 levels in conjunction with a normal TSH. The other immune conditions like ITP and changes in body habitus are new associations, cause of which is yet not identified. ** Photos are available **

## G479(P) ADHERENCE TO BLOOD GLUCOSE MONITORING IN CHILDREN AND YOUNG PEOPLE WITH TYPE 1 DIABETES ON INSULIN PUMP THERAPY IN A TEACHING HOSPITAL

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**Aims** Children and young people with Type 1 diabetes on insulin pump therapy are expected to perform 4–8 capillary blood glucose (BG) tests per day for better glycaemic control. Our objective was to find out whether our patients adhered to the expected BG monitoring.

**Methods** Data was collected from 78 patients during a clinic visit over a period of one year. All children have glucometers which wirelessly transmits the data to their insulin pump. Average numbers of BG tests per day and mean BG levels were downloaded through the pump software for two weeks prior to their clinic visit.

**Results** 48 children (61.5%) did 4–8 BG tests per day while 18 (23.1%) did more than 8 tests. 12 (15.4%) who did less than 4 per day had a mean age of 14.8 years. We found moderately significant negative correlation between age and frequency of BG testing (Pearson's correlation coefficient (R) = –0.57) and also number of BG tests and mean BG levels (R=–0.52). There was a weak negative correlation between number of BG tests and HbA1c levels (R = –0.31). 5 patients (6.4%) entered fictitious BG levels manually into their pump and details are given in the table.