A CASE REPORT OF TRBETA MUTATION LEADING TO RAISED T4 LEVELS

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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 transmit 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.

Conclusion Mutation of the beta thyroid hormone receptor is usually either autosomally 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 **