Aims To describe a case of premature adrenarche with pseudo-hypoparathyroidism, an as yet unreported combination.

Methods An otherwise well 8 year old girl presented to a Paediatric Endocrine Clinic with early pubic hair development suggestive of premature adrenarche. Blood tests revealed hypercalcaemia, elevated phosphate and highly elevated parathyroid hormone (PTH) level, giving a biochemical diagnosis of pseudo-hypoparathyroidism. She had normal stature (height 50th – 75th centile) and no phenotypic features of Albright Hereditary Osteodystrophy (AHO) were identified: obesity, learning difficulties, brachyactyly, short stature, shortened 4th/5th metacarpals, dental hypoplasia or a rounded face.

Results Blood tests revealed low corrected calcium 1.49 mmol/L (reference range 2.2 – 2.7), elevated phosphate 2.78 mmol/L (reference range 0.9 – 1.8) and serum PTH level almost 10 times the upper limit of normal at 66.4 pmol/L (reference range 1.6 – 6.9), with normal Vitamin D 94 nmol/L, normal thyroid function: Free T4 5.4 pmol/L (reference range 12 – 22); TSH 4.8 mIU/L (reference range 0.27 – 4.2).

Management She commenced oral calcium carbonate and alfa-calcidol to correct the severe calcium deficiency and to normalise PTH levels. Progressively increasing doses have been required.

Conclusion Pseudo-hypoparathyroidism is a rare endocrine disorder characterised by resistance to the action of PTH. It has been classified within the AHO group. Recognition of a broader range of phenotypic features and underlying mutations has led to a novel classification system of iPSSD (inactivating PTH/PTHrP signalling disorders) developed by the EuroPHP network. GNAS1 mutations have been identified underlying various pseudo-hypoparathyroidism subtypes, resulting in reduced function of the G-protein coupled to the PTH receptor. G-proteins are also coupled to other hormone receptors; patients with AHO or iPSSD often present with a range of endocrine disorders, for example hypothryoidism. There are cases of individuals with GNAS1 mutations presenting concurrently with precocious puberty and pseudo-hypoparathyroidism but no reported case of premature adrenarche and pseudo-hypoparathyroidism. Awaited genetic studies in our case may be informative.

Aims The National Screening Committee and NICE recommend monitoring of diabetes patients for retinopathy from the age of 12 years as early treatment is known to improve outcomes. Control of HbA1c may reverse background retinopathy and good control of blood pressure can delay diabetic retinopathy by 4 – 5 years, monitoring blood pressure has therefore been included in NICE guidance. We firstly aimed to assess compliance with NICE guidelines at Heartlands Hospital in 2015 – 2016 compared to results from 2011. Secondly we aimed to compare HbA1c levels in those with retinopathy compared to those without.

Methods Review of the electronic database of patients known to the diabetes team at Heartlands Hospital between 1st April 2015 and 31st March 2016. The inclusion criteria were those with type 1 diabetes mellitus and age over 12 years by 31st March 2016.

Results 137 patients met the inclusion criteria of whom 81.6% had been screened for retinopathy, this is compared to 68% in 2011. Of those screened 18% had background retinopathy and 2% preproliferative retinopathy with the remainder normal. Mean HbA1c for those without retinopathy (80.51 mmol/mol) was lower than in those with background retinopathy (91.05 mmol/mol) and preproliferative retinopathy (100 mmol/mol). These HbA1c results were lower than 2011 when mean HbA1c was 83.6 mmol/mol for those without retinopathy and 108.7 mmol/mol in those with background retinopathy. Despite this improvement, HbA1c is higher in our cohort than the national mean. Annual blood pressure check had been performed in 99.3% of patients.

Conclusion Higher HbA1c in those with retinopathy is consistent with the literature. While there has been an improvement in both rates of retinopathy screening and HbA1c levels between 2011 and 2016 at Heartlands hospital, rates of retinopathy overall remain similar. There has however been an improvement in average HbA1c although this is still higher than national average, this may reflect the deprived population served by the hospital. Those who had failed to attend screening were identified to ensure further education.