he and his partner have had a child who is currently on allopurinol for hyperuricemia which is controlling his urate levels.
Discussion: Uromodulin gene related kidney disease Uromodulin gene related kidney disease is an autosomal dominant disease that results in slowly progressive kidney disease characterised by minimal proteinuria without haematuria. Accompanying this is hyperuricemia and gout. It usually presents in early teens progressing to end-stage kidney disease between the fourth and seventh decade. Normalisation of serum urate prevents gout flares but does not alter the course of the CKD. Allopurinol Hypersensitivity syndrome Allopurinol Hypersensitivity is rare (risk estimate 0.1–0.4%), however it carries with it a high mortality rate of up to 25%. Recognised risk factors include pre-existing renal failure and the presence of HLA-B*58:01. The HLA-B*58:01 allele is most common in individuals of Asian descent, with a frequency of 10-15% in the Han Chinese, 12% in Koreans, and 6-8% in individuals of Thai descent . The risk allele is less common among Europeans and Japanese with a frequency of only 1-2%
Key learning points: 1. In young adults presenting with hyperuricemia, gout and CKD consider testing for uromodulin gene related kidney disease. 2. Genetic counselling is important as this is an autosomal dominant condition and early use of allopurinol in off spring will reduce the incidence of gout. 3. Both haemofiltration and Rasburicase resulted in rapid resolution of tophi and healing of tophaceous ulcers but only temporary normalisation of serum urate. 4. The HLAB*58:01 allele is found most commonly in patients with Korean, Han Chinese and Thai descent. These patients should be screened for the HLA-B*58:01 allele and if positive allopurinol should not be commenced. Our patient tolerated Febuxostat without any complications.

26. AUTOSOMAL DOMINANT KIDNEY DISEASE PRESENTING AS TOPHACEOUS, ULCERATING GOUT.

Dr Richard Smith, Salisbury Foundation Trust, Salisbury

Dr Luke Sammut and Dr Sarah Bartram, *University Hospital Southampton*

Introduction: A man in his 30s presented with tophaceous, ulcerating gout and CKD. Investigations confirmed a diagnosis of Uromodulin gene related kidney disease. He was started on Allopurinol but developed a hypersensitivity syndrome. Successful treatment of his gout included Rasburicase followed by Febuxostat.
Case description: A 37 year old male half Pacific Islander and half Singapore (Chinese) presented to Salisbury hospital with a history of tophaceous gout, diabetes and CKD. Examination revealed tophaceous leg ulcers, patellar and olecranon bursae containing tophi and significant joint damage to elbows, ankles and DIPJs all clearly seen on plain radiographs. His eGFR ws 31, Creatinine 217 umol/l, Urea 19.1 mmol/l and urate 779 mmol/l. Allopurinol was commenced but unfortunately he developed a hypersensitivity syndrome and was admitted to ITU with deteriorating renal function, hepatitis and a rash. Haemofiltration was commenced on ITU as treatment for his deteriorating renal function . Whilst on haemofiltration there was noticeable improvement in his leg ulcers. Upon discharge, funding was obtained to commence Rasburicase which resulted in temporary normalisation of his serum urate and significant improvement and healing of the tophaceous leg ulcers. Febuxostat was also commenced and no adverse reaction was observed. His gout remained under good control but because of deteriorating renal function his renal physicians, upon further investigation diagnosed uromodulin gene related kidney disease. With gout under control