showed a high Calcium of 2.89 mmol/L with a low urine Calcium supporting the likely diagnosis of familial hypocalciuric hypercalcaemia. Molecular analysis of AP2S1 gene was abnormal. This result confirmed a molecular diagnosis of FHH type 3. 

**Discussion** FHH is an autosomal dominant condition. FHH1 is caused by loss of function in CASR gene, FHH2 by a mutation in GNA11 gene and FHH3 identified as mutation in AP2S1 gene. Mutations lead to an elevation of the normal set point for maintaining normal plasma calcium levels. This causes mild to moderate hypercalcaemia with ionised calcium levels usually within 10% of the upper limit of normal. Urine calcium/creatinine ratio tends to be low in FHH whereas it is usually elevated in primary hyperparathyroidism. In the classic description of FHH, individuals are asymptomatic with normal bone density and no intervention is usually required.

**Presentation** A healthy caucasian 3 year old girl was referred due to bowing of her femora, apparent since she started walking at 13 months. She was reported to be clumsy and tire easily. There was no history of fractures or leg pain. Her height had dropped from the 75th centile to between the 25th and 50th centile. Initial investigations showed mildly low corrected calcium and phosphate, slightly raised alkaline phosphatase and a sufficient vitamin D level of 50nmol/L. She was treated with oral Vitamin D supplements. X-ray of knees was normal. On review after 6 months, bowing had progressed and height had fallen further. This led to the consideration of rarer forms of rickets.

**Further investigations** Urinary calcium creatinine ratio was normal at 0.07 but urinary phosphate: creatinine ratio was elevated at 4.36; with reduced tubular reabsorption of phosphate - in keeping with a diagnosis of X-linked hypophosphataemic rickets. This was confirmed by detection of a mutation in the PHEX gene.

Skeletal survey showed lower limb abnormalities, including flared metaphyses, widening of the growth plates and buttressing of the femur and tibia. Renal ultrasound showed no nephrocalcinosis. Parental blood tests showed a slightly low calcium and phosphate, slightly raised alkaline phosphatase and a sufficient vitamin D level of 50nmol/L. She was treated with oral phosphate supplements and alfacalcidol. Since commencing treatment her serum phosphate levels usually within 10% of the upper limit of normal. Urine calcium/creatinine ratio tends to be low in FHH whereas it is usually elevated in primary hyperparathyroidism. In the classic description of FHH, individuals are asymptomatic with normal bone density and no intervention is usually required.

**Progress and treatment** She was treated with oral phosphate supplements and alfalcacidol. Since commencing treatment her growth has improved and she remains active but still tires easily. She was subsequently diagnosed with moderate sensorineural hearing loss, another feature of hypophosphataemic rickets, and has had bilateral hearing aids fitted.

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