

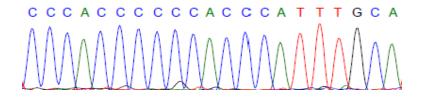
Appendix 2

This appendix was part of the submitted manuscript and has been peer reviewed. It is posted as supplied by the authors.

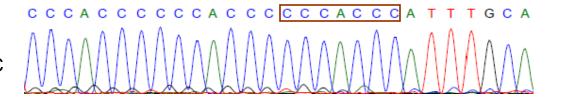
Appendix to: Ito N, Peña AS, Perano S, et al. First Australian report of vitamin D-dependent rickets type I. *Med J Aust* 2014; 201: 420-421. doi: 10.5694/mja13.00220.

Appendix 2: genomic DNA analysis of CYP27B1 in the index case's family

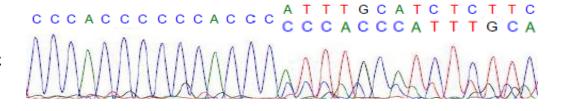
Reference Sequence



Index Case (Female 2y) homozygote c.1325-1326insCCCACCC



Father 37y heterozygote c.1325-1326insCCCACCC



Mother 37y heterozygote c.1325-1326insCCCACCC

