

C.194 a>C (Q65P) mutation in the LMX1B gene in patients with nail-patella syndrome associated with glaucoma

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Purpose: To report the clinical, ophthalmic, extraophthalmic, and genetic characteristics of nail-patella syndrome (NPS) in a Chilean family and to investigate the expressivity of open angle glaucoma (OAG) and ocular hypertension (OHT) in the family members. **Methods:** Five family members affected with NPS and two unaffected members underwent a complete ophthalmologic examination, including computerized visual field, optical coherence tomography (OCT) of the optic disc and ultrasound pachymetry. Renal function was assessed by urinalysis and blood tests. Orthopedic evaluations were also performed, including radiological studies of the wrist, elbow and hip joints. Genomic DNA was extracted from peripheral leukocytes of the five affected and two unaffected family members. Exons 2-6 of the LIM homeobox transcription factor 1-beta (LMX1B) gene were screened for mutations by DNA sequencing of the proband. We also screened for mutations in exon 2 by polymerase chain reaction-restriction fragmen