# INF2 gene mutation, p.Leu77Pro variant causing Charcot Marie Tooth disease; A case report

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**Case Diagnosis:**

Charcot-Marie-Tooth Disease with Focal Segmental Glomerulosclerosis (FSGS)

**Case Description:**

A twelve-year-old, Hispanic male with a past medical history of bilateral foot drop, renal transplant secondary to focal segmental glomerulosclerosis (FSGS) presented to Neuromuscular Clinic with progressive weakness and hypoesthesia in his bilateral lower extremities over 2 months. He was referred to the Neuromuscular clinic by his nephrologist for lower extremity hypoesthesia, after tacrolimus toxicity was ruled out for transplant rejection prophylaxis. Notable on the physical exam was bilateral foot drop, normal foot arches and hypoesthesia in bilateral feet, with steppage gait. Both parents were present on examination and have no significant family history of neuromuscular disease and normal foot arches. Upon obtaining genetic testing results, the patient was found to have a heterozygous Inverted formin-2 (INF2) gene mutation with a p.Leu77Pro variant. The patient and family were educated on prognosis, management with braces and therapies and follow up in neuromuscular clinic.

**Discussion:**

CMT encompasses a group of disorders called hereditary sensory and motor neuropathies and is the most common neuromuscular disorder that damage the peripheral nerves. Patients typically present with progressive distal-muscle weakness and atrophy, reduced tendon reflexes, and foot deformities.[1] Over 40 different genes have been associated with CMT.[2] Although autosomal dominant Charcot–Marie–Tooth type 1 is the most prevalent form.[3-4] After literature review, there are no case reports reporting an INF2 mutation regarding the p.Leu77Pro variant. The INF2 genetic mutation shows a strong association with FSGS and patients of Hispanic descent. Clinically, 50% of cases with this mutation are sporadic and present from 5-28 years old. Which includes proteinuria, as well as progressive to end stage renal disease to the constellation of CMT symptoms.

**Conclusion:**

While the INF2, p.Leu77Pro gene mutation is an unusual cause of CMT. The presentation of FSGS, and bilateral foot drop in a pediatric patient, should be diagnostically investigated for CMT.

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