

Annual Congress on

# Rare Diseases & Orphan Drugs

October 26-27, 2016 Chicago, USA

## An online survey of neurologists about Charcot-Marie-Tooth disease type 1A

**Xavier Paoli**  
Pharnext, France

Charcot-Marie-Tooth (CMT) disease is the most common inherited disorder of the peripheral nervous system. CMT type 1A (CMT1A) is an autosomal dominant and demyelinating subtype of CMT, classified as an orphan disease with around 125000 patients across Europe and the US. Presently incurable, CMT1A significantly impairs quality of life of affected patients. Our survey aimed at understanding the CMT1A patient journey across the US and European healthcare systems and identifying potential barriers that could be alleviated. In October 2014, an online questionnaire was administered to 128 European and US neurologists. Altogether, respondents followed a pool of over 2000 CMT1A patients and had mainly hospital-based activities. Neurologists declared that adult patients usually seek medical care due to symptoms alone contrary to children where family history seems to be more decisive. For most patients, general practitioners, pediatricians and neurologists are the first physicians visited before reaching CMT experts. Physicians use clinical symptomatology, electrophysiology and genetic testing as primary diagnosis tools. Nearly 70% of patients seem to get diagnosed within two years after first symptoms and before the age of 30. Most neurologists surveyed use primarily clinical evaluation of symptoms to gauge disease severity. More than 80% of their patients suffer from mild to moderate CMT1A. Physical therapy, orthotics and pharmacotherapy for neuropathic pain are the most frequent supportive cares. This survey offers a better understanding of the current CMT1A patient journey where the flow could be improved through disease awareness programs developed and led by both neurologists and patient organizations.

### Biography

Xavier Paoli is currently the Director of Commercialization Strategy at Pharnext. He is In-Charge of developing the optimal commercialization plan for all Pharnext products. Prior to joining Pharnext, he has held various marketing/brand management positions in biopharmaceutical/pharma companies such as GlaxoSmithKline, UCB Pharma and Alexion Pharmaceuticals. He holds a Master's degree in Marketing from the HEC Paris Business School and a Master of Science in Genetics and Immunology from the Science University Denis Diderot in Paris.

xpaoli@pharnext.com

Notes: