Variant	Prediction on protein	Inheritance	Segregation	Phenotype	Method	Comments
Pathogenic or likely pathogenic variants						
c.-84-1G>C	p.?	sporadic	de novo	WS4 or PCWH (learning delay, hypotonia at birth; short segment HSCR)	Sanger	
c.44_62del	p.(Val15Alafs*11)	?	ND	WS4	Sanger	Sun, *Sci Rep* 2016;6:35499
c.61del	p.(Arg21Alafs*11)	sporadic	parents not carriers	PCW + olfactory bulbs agenesis	Sanger	
c.89C>A	p.(Ser30*)	familial	cosegregates	PCWH in index case and mother, isolated SNHL in sister	Sanger	Cassatella, *Eur J Endocrinol* 2018;178:377
c.236T>G	p.(Val79Gly)	familial	de novo in father	PCWH (index case : demyelinating neuropathy, SNHL ; son : peripheral neuropathy + HSCR + depigmentation, hearing is said to be normal)	Sanger	
c.255G>A	p.(Trp85*)	sporadic	de novo	WS4 (long segment HSCR)	Sanger	Sun, *Sci Rep 2016;*6:35498
c.325A>T	p.(Asn109Tyr)	sporadic	de novo	WS2 + anosmia	Sanger	
c.331T>C	p.(Phe111Leu)	sporadic	ND	WS2 or PCW (motor delay and ID), anosmia and hypoplastic olfactory bulbs	Sanger	Liu, *Int J Pediatr Otorhinolaryngol 2019;*130:109806
c.333C>A	p.(Phe111Leu)	sporadic	parents not carriers	WS2	NGS panel	
c.335T>C	p.(Met112Thr)	sporadic	de novo	WS2 + vestibular areflexia	NGS panel	
c.335T>C	p.(Met112Thr)	familial	de novo in mother	WS4 in 2 index case brothers (short segment HSCR), WS2 in mother	Sanger	
c.335_336del	p.(Met112Serfs*21)	sporadic	parents not carriers	WS2 + vestibular areflexia	Sanger	
c.338T>C	p.(Val113Ala)	familial	cosegregates	WS4 in 2 index case brothers , one has a possible hypogonadism; a third brother has SNHL; father has WS2	NGS panel	
c.341G>A	p.(Trp114*)	sporadic	parents not carriers	WS4	NGS panel	
c.342G>A	p.(Trp114*)	familial	?	WS2	sanger	
c.342G>T	p.(Trp114Cys)	sporadic	de novo	WS2 + mild hypotonia	sanger	
c.355C>G	p.(Arg119Gly)	sporadic	parents not carriers	WS2	Sanger	
c.356_357del	p.(Arg119Glnfs*14)	sporadic	parents not carriers	WS2 or PCW, hyperpigmentation of hands	Sanger	