

TABLE I. Summary of clinical findings.

<b>Patient</b>	<b>1</b>	<b>2</b>	<b>3</b>	<b>4</b>
<b>Age at last examination</b>	7 y	4 y	45 y	15 y
<b>Sex</b>	M	M	F	F
<b>IUGR</b>	-	+	-	-
<b>Birth at term</b>	+	+	+	+
<b>Gastro-intestine</b>				
Meconium emission retardation	-	-	?	-
Constipation	+	+	-	+
Hirschprung disease	+ (long-Type)	-	-	-
Other	-	-	-	Achalasia
<b>Neurological / Development</b>				
Neonatal hypotonia	+	+	?	-
Age at sitting	10 m	> 10 m	?	10 m
Age at walking	18 m	NA	2 y	2 y
Muscle weakness in infancy	+	-	-	-
Muscle weakness in adulthood	NA	NA	-	-
Speech delay	+	+	+	+
Motor developmental delay	+	+	+	+
Mental retardation	-	+	-	-
<b>Hearing loss / Inner ear imaging</b>				
Hearing loss	+	+	+	+
Type	Profound, bilateral	Profound, bilateral	Profound, bilateral	Profound, bilateral
Bilateral	+	+	+	+
Abnormal semicircular canal	-	+	+	+
		+	?	?
<b>Vestibular dysfunction</b>	-	?	-	+?
<b>Pigmentation defects</b>				
Eyes				
Bright blue irises	+	+	+	+
Iris heterochromia	-	-	-	-
Skin				
Depigmented areas	+	+	+	+
Café au lait spots	+	-	-	-
Freckles	-	-	+	-
Hair				
White forelock	+	+	+	+
Premature graying (age)	-	+ (9 m)	+ (16 y)	+ (12 y)
Eyebrows premature graying(age)	-	+ (9 m)	+ (16 y)	+ (12 y)
<b>Eye anomalies</b>				
Telecanthus	?	+	-	-
Retinal pigmentation defect	?	-	+	+
Nystagmus				
During first month	+	+	?	-
		+	?	+
At 3 months	-	NA	?	+
At 2 years	NA	NA	-	-
As adult				
<b>Abnormal olfaction</b>	?	NA	-	-
<b>CT Scan</b>				Absence right OB
<b>Teeth</b>				
Late dental eruption	NA	?	?	+
Caries	NA	NA	+	+
<b>Genitalia</b>	Neonatal ectopic testes, resolved spontaneously	Normal male	Normal female	Normal femalebut delayed puberty
<b>Other</b>				Pituitary insufficiency (GH and gonadotropic)
<b>Genetic</b>				

<i>SOX10</i> mutation	c.336G>A; p.Met112Ile	c.336G>A ; p.Met112Ile	c.336G>C p.Met112Ile	c.336G>C p.Met112Ile
rs2435357 ( <i>RET</i> intronic)	C/C	C/C	C/C	C/T

NA, Not applicable; y, year;; m, month; ?, not explored or unknown; OB, olfactory bulb