

<b>Proband ID</b>	<b>Family history</b>	<b>Hearing loss</b>	<b>Other features</b>
P794	Spor	Progressive bilateral, mixed HL; SNHL component profound	External, middle and inner ear anomalies, myopia, bilateral retinal coloboma, hypothyroidism, coeliac disease
P152	Spor	Moderate bilateral SNHL	Global developmental delay, seizures, microcephaly, dysplastic facial features, fingers and toes syndactyly
P045	Fam	Severe bilateral SNHL	Unilateral heterochromia, dorsiflexion of 2nd toes
P584	Spor	Mild to moderate bilateral SNHL	Delayed motoric milestones, difficulties in walking, progressive distal weakness of the lower and upper limbs, cerebellar dysfunction, peripheral motor and sensory neuropathy
P144	Spor	Mild bilateral SNHL	Congenital arthrogyrosis, delayed motoric milestones, severe scoliosis
P552	Spor	Moderately severe bilateral conductive HL	Malar hypoplasia, micrognathia, thumb hypoplasia, radioulnar synostosis
P555	Fam	Moderately severe bilateral SNHL	Both parents are hearing impaired, father also has RP along with some of his other close relatives
P314	Spor	Moderately severe bilateral SNHL	Microphthalmia, solitary kidney, omphalocele, dysplastic facial features; normal growth and development
P852	Fam	Moderate bilateral SNHL	Retinitis pigmentosa
P506	Fam	Severe bilateral SNHL	Severe developmental delay, cerebellar hypoplasia (vermis and hemispheres)
P654	Spor	Severe bilateral SNHL	Intestinal atresia type IIIa, unilateral hydronephrosis, developmental delay
P554	Spor	Moderate bilateral SNHL	Intellectual disability, autism, seizures, scoliosis, bronchogenic cyst of the neck
P052	Spor	Moderately severe bilateral conductive and SNHL	Aural atresia
P652	Spor	Moderate unilateral conductive and SNHL	Developmental delay, dysplastic facial features, choroidal coloboma, amblyopia, Dandy Walker malformation
P352	Fam	Profound bilateral SNHL	Hypopigmentation of hair
P952	Spor	Moderate unilateral conductive HL	Developmental delay, on the right side microtia (third degree), anophthalmia, and facial nerve paralysis, agenesis of corpus callosum
P752	Spor	Severe bilateral SNHL	Developmental delay, tracheoesophageal fistula, synostosis of the 5th and 6 <sup>th</sup> rib on the right side, unilateral cryptorchidism