



## Autism and Deafness, A Diagnostic Clinical Challenge

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### Abstract

Autism Spectrum Disorder (ASD) and congenital deafness frequently co-occur, with higher rates of ASD reported in deaf children than in the general population. Overlapping clinical features often complicate differential diagnosis and may lead to diagnostic delay, with significant consequences for early intervention and family counselling. We report the case of an 8-year-old boy with congenital bilateral sensorineural hearing loss diagnosed in early infancy, who later developed social communication regression and was diagnosed with moderate-to-severe ASD. The patient underwent early auditory rehabilitation and bilateral cochlear implantation, showing good functional adaptation despite the neurodevelopmental diagnosis. Whole-exome sequencing subsequently identified a homozygous pathogenic variant in MYO15A (c.4107C>A; p. Ser1369Arg), consistent with autosomal recessive non syndromic hearing loss (DFNB3), as well as heterozygous variants of uncertain significance in CHD8 and SHANK2, genes recognized as autism susceptibility genes. This case illustrates a complex neurodevelopmental phenotype with a monogenic cause of deafness and additional genetic susceptibility potentially contributing to ASD through a multifactorial or oligogenic mechanism. Early recognition of ASD in deaf children is challenging but essential to ensure timely multidisciplinary intervention, realistic expectations regarding cochlear implantation outcomes, and appropriate genetic counselling. Comprehensive genetic evaluation can play a key role in clarifying diagnosis and guiding clinical management in children presenting with both deafness and ASD.

**Keywords:** Autism spectrum disorder; Diagnosis

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### Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder characterized by core deficits in social communication/interaction and presence of restrictive/repetitive patterns of behaviour.

The rates of ASD in deaf children are higher than in the general population and early diagnosis of the coexistence of both can be challenging, because they are differential diagnosis of each other. Diagnostic delays are expected and have a severe impact for the child and family.

We present an 8-year-old male, diagnosed at 7 months of age with congenital neurosensory bilateral deafness and at 4 years diagnosed with ASD. After extensive etiologic investigations, initially negative, the exome revealed three different variants in distinct genes, two associated with ASD susceptibility and one causing deafness, each contributing to the phenotype.

Exome sequencing findings in autism susceptibility genes may reflect two independent Mendelian conditions or a two-hit or oligogenic mechanism, with modification of expressivity on one another. It is essential to recognize both diagnoses and genetic susceptibilities to provide a more accurate genetic counselling to the patient and family.

### Case Presentation

An 8-year-old boy was referred for evaluation because of bilateral congenital deafness and autism spectrum disorder.

Pregnancy was term and planned. The mother had antiphospholipid syndrome, treated with aspirin and heparin. Birth weight and length were appropriate for gestational age. The mother had three previous miscarriages two in the first trimester and one at approximately five months due to a fetus with multiple congenital anomalies. In this pregnancy, neonatal hydronephrosis was

diagnosed on prenatal ultrasound, and the child had prophylactic antibiotics for four months to prevent urinary tract infections. The remainder of the neonatal period was unremarkable.

The patient was diagnosed in the first months of life with congenital neurosensorial bilateral deafness, and he started rehabilitation. He had a speech and language and occupational therapy evaluation at 10 months of age, which revealed a motor delay (not able to sit without support), but an otherwise normal developmental assessment including typical social interaction. He experienced recurrent otitis media in early infancy and underwent planned otolaryngologic surgery at 15 months of age. The patient had the first cochlear implantation performed at 18 months of age, followed by regular otolaryngology follow-up and auditory-verbal and speech-language rehabilitation. He received the Measles Mumps Rubella (MMR) vaccine one week later. Varicella infection occurred at 16 months.

Since 15-18 months a social communication regression was observed, with loss of eye contact and reduced social engagement. Stereotyped movements increased in frequency and occurred during states of excitement. He lost previously acquired receptive signing and developed sensory processing difficulties and lack of symbolic play, with repetitive activities and inconsistent responses to sound.

With 2,5 years of age, he had the diagnosis of moderate-to-severe autism spectrum disorder with significant impairment in communication, socialisation and behaviour, and episodes of aggression.

He presented with sleep difficulties that were treated with melatonin without benefit, and subsequently developed self- and hetero-aggressive behaviour, which was managed with risperidone. Regarding hearing, he had a second cochlear implant at 8-years old with good adaptation and results.

Family history was notable for autoimmune disease in the mother (thyroiditis, antiphospholipid syndrome and lupus) and maternal grandmother (vitiligo, thyroiditis and chronic urticaria). A paternal cousin had Waardenburg syndrome, and another cousin had ASD. A 5<sup>th</sup> degree cousin had deafness. No other relevant history was reported.

On physical examination, he had prominent and protruding ears, congenital hypopigmented, serpiginous patch over the left infraclavicular region and three café-au-lait macules. No other findings on physical examination.

He had several etiological investigations, including cytomegalovirus testing on the new born dried blood spot and urine, that was negative, metabolic screening included biotinidase activity and creatine studies that were unremarkable. Genetic testing for GJB2/GJB6 (connexin 26/30) detected no pathogenic variants, excluding the most common genetic cause of autosomal recessive non-syndromic sensorineural hearing loss. Fragile X testing and karyotype were normal, FISH for 22q11.2 deletion (DiGeorge syndrome) was negative and chromosomal microarray (array-CGH) was normal. Electroencephalogram and MRI of the brain and inner ear (including cochlea) were unremarkable. Abdominal ultrasound, cardiology and ophthalmology assessments were normal.

In 2018 a whole-exome sequencing was ordered, and it identified a homozygous pathogenic variant in MYO15A, c.4107C>A, p. Ser1369Arg (exon 9), consistent with autosomal recessive non

syndromic hearing loss (DFNB3), was detected and explaining the clinical picture of deafness. Additionally, variants of uncertain significance in CHD8 (c.5902C>T, p. His1968Tyr (exon 31)), which has not been previously reported, and in SHANK2 (c.3031C>G, p. Arg1011Gly (exon 15)) were identified. These genes are involved in the period of early neurodevelopment and described as ASD susceptibility genes. As ASD has a known multifactorial aetiology, the simultaneous presence of these variants may be interpreted as contributory to the neurodevelopmental phenotype.

The patient and family were referred to Genetics counselling.

## Discussion

Deafness and ASD are highly co-occurrent and rates of ASD in deaf children (7% to 9%) are higher than in the general population (1,7% to 2%). This may be due to common underlying etiologies, such as prematurity, congenital infection or genetic syndromes [1,2].

Although controversial, studies have shown an increased prevalence of central and peripheral hearing loss in people with ASD. The relationship between autism and hearing loss is thought to be better explained by the presence of neurological alterations that lead to the development of these two conditions rather than that hearing loss causing the development of ASD [1].

Early diagnosis of the coexistence of these disorders is often challenging, because of overlapping symptoms and uncertainty if it results from social or communication problems, and appropriate early diagnosis is mandatory [3]. Diagnosis of ASD in deaf children happens on average 3 years later than in hearing children, with average ages of diagnosis of 66,5-76 months in deaf children and 38 months in hearing children. This can be related to the lack of validated gold-standard assessments for ASD in deaf children, complicated differential diagnosis and lack of clinician expertise. The diagnostic delay can lead to delays in accessing appropriate early intervention [2].

We emphasise that despite the coexistence of both diagnoses, in this case, the ASD diagnosis was not severely delayed due to the deafness.

It is also important because it allows families to have realistic expectations regarding recommended cochlear implantation, as language and communication skills in children with ASD have a neurological origin, and so it is expected that the development of language skills does not have the same progress as for deaf children, because children with ASD seem to have a significant difference in auditory perception and verbal clarity when compared to deaf but neurotypical children [3].

Despite the previously described difficulties of effectively using a cochlear implant, our patient had an unusually good adaptation with both of his implants, even after ASD diagnosis.

The variants found in this patient are associated with ASD but they are still considered Variants of Uncertain Significance (VUS), and authors emphasize that these genetic variants should be seen as susceptibility genes in a multifactorial disease, where they could contribute to the ASD phenotype.

The SHANK2 is a post-synaptic scaffolding protein that anchors a protein complex and has functions in early neuronal development and in glutamatergic synapses, being a suppressor of dendrite branching. SHANK2 mutations can cause hyperconnectivity of human neurons

and increased dendritic complexity which are thought to underlie ASD [4-11].

CHD8 is transcriptional regulator protein that is implicated in cell cycle, cell adhesion, neuronal development, myelination and synaptogenesis. Pathogenic variants in CHD8 are amongst the most common genetic variants associated with ASD, with a broad phenotypic spectrum [4-8].

MYO15A encodes myosin XVa, a protein required for development and maintenance (elongation and mechanotransduction) of sensory stereocilia in the inner ear hair cells. Pathogenic variants in this gene are a common cause of autosomal recessive nonsyndromic hearing loss. Clinical spectrum is extensive, with severity being related to the nature and location of the variant [6]. The accurate diagnosis is relevant for the child and family, as it allowed the diagnostic odyssey to end and allowed necessary genetic counselling for the family.

## Conclusion

This case highlights a dual diagnosis in an adolescent with congenital sensorineural hearing loss and autism spectrum disorder: a homozygous pathogenic MYO15A variant consistent with DFNB3, together with heterozygous variants in CHD8 and SHANK2, possibly contributing to the neurodevelopmental phenotype of ASD. In children presenting with ASD and deafness, comprehensive exome sequencing can aid in clarifying diagnosis of the phenotype.

This case report also highlights the importance of recognizing and diagnosing ASD in deaf children with referral to early intervention services. Early recognition of dual diagnoses in this case also allowed the family to have realistic expectations regarding communication outcomes after cochlear implantation and allows for timely multidisciplinary care including early referral for therapies.

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