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REPORT OF A PEDIATRIC PATIENT WITH A RARE SYNDROME DDX3X PRESENTING TO AUDIOLOGY, WITH A BRIEF REVIEW OF THE LITERATURE

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Abstract

Background:
DDX3X, is a very rare syndrome presenting in Pakistan for the first time. It is due to the second most frequently mutated gene in Neuro developmental delays, accounting for 0.37% of all de novo variants (prevalence of 3.6/100,000). This genetic condition that occurs predominantly in females, and very rarely in males.

Purpose:
The purpose of our study was to assess and report on hearing threshold estimations using objective methods of hearing evaluation. This was a unique opportunity for an audiology department to be part of a multi-disciplinary team for estimating hearing thresholds which form an integral part of the diagnosis of DDX3X.

Research design:
This study is a case report of hearing threshold estimation in a child diagnosed with a DDX3X Syndrome employing objective tests of hearing with the child under oral sedation, oral chloral hydrate (50-60 mg/kg).

Study sample:
A child 2.4 years old, female, second of the two, born to parents with normal hearing was referred to us. The mother denied prematurity, low birth weight, jaundice requiring blood transfusion, consanguinity, ECMO, history of new born hearing screening and NICU stay more than 5 days.

Method:
Otoacoustic emissions was performed by using Interacoustics to evaluate the function of outer hair cells, tympanometry was performed to evaluate any middle ear analysis. Auditory Brainstem Response Audiometry(ABR) was performed to evaluate hearing threshold of the patient.

Results:
Otoacoustic emissions of patient were bilateral Pass, suggesting hearing threshold levels better than 35dBnHL. Tympanometry showed Type A tympanogram suggesting no mid ear pathology. ERA Wave V was traced down to 30 dBnHL in both the ear (clear response) suggesting normal hearing threshold levels with a CR in both ears.

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Conclusion:

Our study showed that child with DDX3X Syndrome had normal hearing threshold levels. We advised 6 monthly yearly follow to monitor hearing threshold for the next 2 years. Referral to speech pathology as part of the multi-disciplinary team was also offered. Detailed behavioral and developmental assessment of the child was already undergoing in other specialties of the team.

Introduction:

DDX3X syndrome, also referred to as DDX3X-related neurodevelopmental disorder (MIM #300958), is an ultra-rare X-linked disorder being reported here for the first time from Pakistan. The condition is caused by pathogenic variants in DDX3X, a DEAD-box ATP-dependent RNA helicase gene central to translation regulation, mRNA metabolism, stress-granule assembly, and neurogenesis (1). Since the syndrome was first delineated in 2015 by Snijders Blok et al., who identified de novo DDX3X variants in females with unexplained intellectual disability, more than 200 individuals have now been described worldwide (2,3). Pathogenic DDX3X variants account for an estimated 1-3% of unexplained intellectual disability in females and approximately 0.37% of all de novo variants identified in neurodevelopmental disorders overall, corresponding to an estimated prevalence of 3.6 per 100,000 (4).

DDX3X escapes X-inactivation, so affected females are functionally heterozygous for pathogenic variants, which largely accounts for why the disorder disproportionately affects females; hemizygous males with de novo loss-of-function variants are presumed not to survive to term, a finding also supported by murine models (5). Nonetheless, DDX3X-related neurodevelopmental disorder in males is increasingly recognized. A 2025 multicenter study describing 19 affected males - the largest male cohort reported to date - found that surviving males almost exclusively carry missense variants, including two recurrent hotspot substitutions, with truncating variants presumed embryonically lethal; the male phenotype spans intellectual disability, speech and language delay, behavioral difficulties, and structural brain anomalies, and is generally milder than, but overlapping with, the female phenotype (6).

The behavioral and neurodevelopmental phenotype remains the most consistent feature of the syndrome, encompassing intellectual disability, autism spectrum disorder, attention-deficit/hyperactivity disorder, anxiety, self-injurious behavior, and expressive language impairment severe enough that around half of affected girls remain nonverbal beyond five years of age. Since 2022-2023, larger natural history and genotype-phenotype studies have substantially refined the clinical picture of DDX3X syndrome. Levy et al.'s multi-cohort synthesis established consensus surveillance recommendations and identified auditory abnormalities - both sensorineural and conductive hearing loss - in approximately 8% of affected individuals (13 of 160), alongside high rates of intellectual disability, hypotonia, gastrointestinal dysfunction, ophthalmologic anomalies, and abnormal brain MRI, most commonly corpus callosum abnormalities (7). Building on this, a large multicenter genotype-phenotype study posted in late 2025 - the second-largest DDX3X cohort assembled to date, comprising 52 females and 7 males - used in silico protein-folding analysis to demonstrate, for the first time, a quantitative correlation between the degree of DDX3X monomer misfolding and phenotypic severity, together with associations between variant type/location and specific features such as growth abnormalities, epilepsy, brain anomalies, and motor findings (8).

Most recently, a 2026 study applying AI-assisted facial analysis (Face2Gene) proposed a recognizable facial gestalt for DDX3X syndrome for the first time, describing a consistent pattern of thin upper lip, flat philtrum, broad nasal bridge, and upslanting palpebral fissures, together with previously under-recognized features including long uniform eyebrows, tapered chin, short neck, and proximal thumb placement, with the trained

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algorithm correctly flagging the syndrome in 92% of cases (4). A 2025 review has additionally highlighted emerging findings in the inflammatory cytokine profile of DDX3X syndrome, raising the possibility of future disease-modifying or precision-targeted therapeutic approaches. Individuals with DDX3X syndrome are also increasingly recognized as having relatively preserved social engagement compared with idiopathic autism spectrum disorder, a distinguishing feature that may aid clinical differentiation (9).

Against this backdrop of rapidly expanding genotype-phenotype knowledge, the auditory phenotype of DDX3X syndrome remains comparatively under-characterized. Although Levy et al. reported an 8% prevalence of hearing loss and recommended universal hearing screening with audiology referral for suspected cases, and patient-registry data describe conductive hearing loss and recurrent otitis media in a substantial minority of affected males (10), detailed objective audiological test batteries - otoacoustic emissions, tympanometry, and auditory brainstem response performed together in a single patient - have rarely, if ever, been reported for this syndrome. Hearing loss in early childhood, irrespective of cause, carries well-documented consequences for speech-language development, education, cognition, and quality of life (11), a concern of particular relevance in a population already vulnerable to communication delay from the primary neurodevelopmental disorder. Given the global burden of hearing loss - the WHO estimates that over 1.5 billion people currently live with some degree of hearing loss, projected to reach 2.45 billion by 2050 (12,13) . Given that 1 to 3 infants per 1000 live births are born with hearing loss (14). The objective audiological evaluation should logically form a routine component of the multidisciplinary work-up for any child with a genetic neurodevelopmental syndrome.

We therefore present the first reported case of DDX3X syndrome from Pakistan, with the aim of providing a detailed, objective audiological characterization - using otoacoustic emissions, tympanometry, and auditory brainstem response performed under sedation - of a child with genetically confirmed DDX3X syndrome. Our goal is to contribute case-level audiological data to a literature that, despite considerable recent expansion in genotype-phenotype and neurobehavioral characterization, remains comparatively sparse on the auditory phenotype, and to underscore the role of audiology within multidisciplinary care pathways for rare neurodevelopmental syndromes in low-resource settings.

Material and methods:

A child 2.4 years old, female, second child born to parents with normal hearing came in Audiology Centre, Lahore for detailed audiological assessment and middle ear evaluation. The parents gave their consent for sedation and procedure offered. The mother denies prematurity, low birth weight, jaundice requiring blood transfusion, consanguinity, ECMO, history of new born hearing screening and NICU stay more than 5 days. Their first sibling was normal in all developmental milestones.

The child was clinically diagnosed with rare de novo DDX3X syndrome. The child had history of globally delayed developmental milestones and normal birth history. The child had complaint of speech delay. There was no family history of hearing loss.

We performed otoscopy, otoacoustic emissions, tympanometry and auditory brainstem response audiometry for hearing and middle ear evaluation. On physical examination, both the external ear canal and tympanic membrane appeared normal.

Tympanometry was performed by Interacoustics Titan to evaluate any middle ear pathology. The types of tympanogram are given in table 1. Type A tympanogram specifies normal middle ear function. Type As specifies reduced compliance of tympanic membrane. Type Ad specifies high compliance. Type B has flat tympanogram with normal volume suggesting otitis media with effusion. Type C has reduced middle ear pressure suggesting Eustachian tube dysfunction.

Otoacoustic emissions were performed by using Otoread by Interacoustics. The result of OAEs is given as pass or refer. Pass suggest hearing threshold levels are better than 35 dBnHL. Refer

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suggests hearing threshold levels are worse than 35dBnHL. Pass criteria was set as 3 frequencies pass. SNR of 4dB was set. Average time was 64s. 1.5 to 4 kHz frequencies were tested.

ABR was conducted by Interacoustics Eclipse to evaluated hearing threshold levels of the patient. The parents gave their consent for sedation and procedure offered. The summary of ABR parameters used are given in table 2.

The test guidelines were followed in audiology center, Lahore. The tests were conducted in a sound treated room. Alcohol swabs and abrasive cream was used to clean the skin of the patient for electrode placement. The impedance of the electrodes was 0.5kOhm.

In our study, Alternating CE-Chirp stimulus was presented at the rate of 44.1 per second with 15 ms time window. Low pass filter of 50 Hz was used and high pass filter of 1500Hz was used. Wave V was identified. Positive electrode was placed on forehead, common electrode a placed on above eyebrow and negative electrode were placed on mastoid. ABR was run at intensity of 30dBnHL, 40dBnHL and 50dBnHL. Informed consent was taken from the patient.

Results:

Tympanometry revealed Type A tympanogram in both ears, which signifies normal middle ear pressure and function, as shown in figure 1. The volume of right and left ear canal was 1.0 ml. the pressure of right and left middle ear was -3daPa and -107 daPa respectively. These values fall within normal pediatric range according to British Society of Audiology. The compliance of tympanic membrane was 0.27ml and 0.34ml respectively. The normal values specified by BSA are shown in table 1:

Transient evoked Otoacoustic emissions of patient were Pass in both ears, suggesting hearing threshold levels were better than 35dBnHL in both ears. It is represented as figure 2-A and 2-B.

In ABR Wave V was traced at 30dBnHL in both ear, suggesting normal hearing threshold levels, as shown in figure 3. Figure 4 and 5 represents Fmp curve and recorded parameters respectively. In right ear wave reproducibility was 95% and in left ear the wave reproducibility was 87%. Fmp was recorded as 10 at right ear and 6.3 in left ear showing clear response with residual noise as 25.8nV and 22.3 nV in right and left ear respectively.

Figure 1: Tympanogram

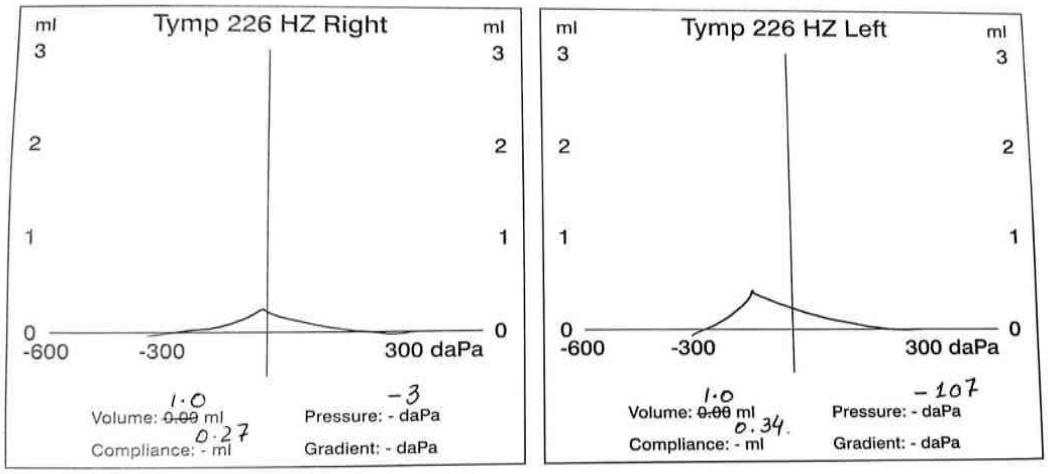


Table 1: Normal values of tympanogram

	Adults	Children
Volume	0.6 to 2.5 cm^3	0.4 to 1.0cm^3
Pressure	+50 to -50daPa	+50 to -150daPa

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Compliance	0.3 to 1.6cm^3	0.2 to 1.6cm^3

Figure 2-A: Left Ear OAEs

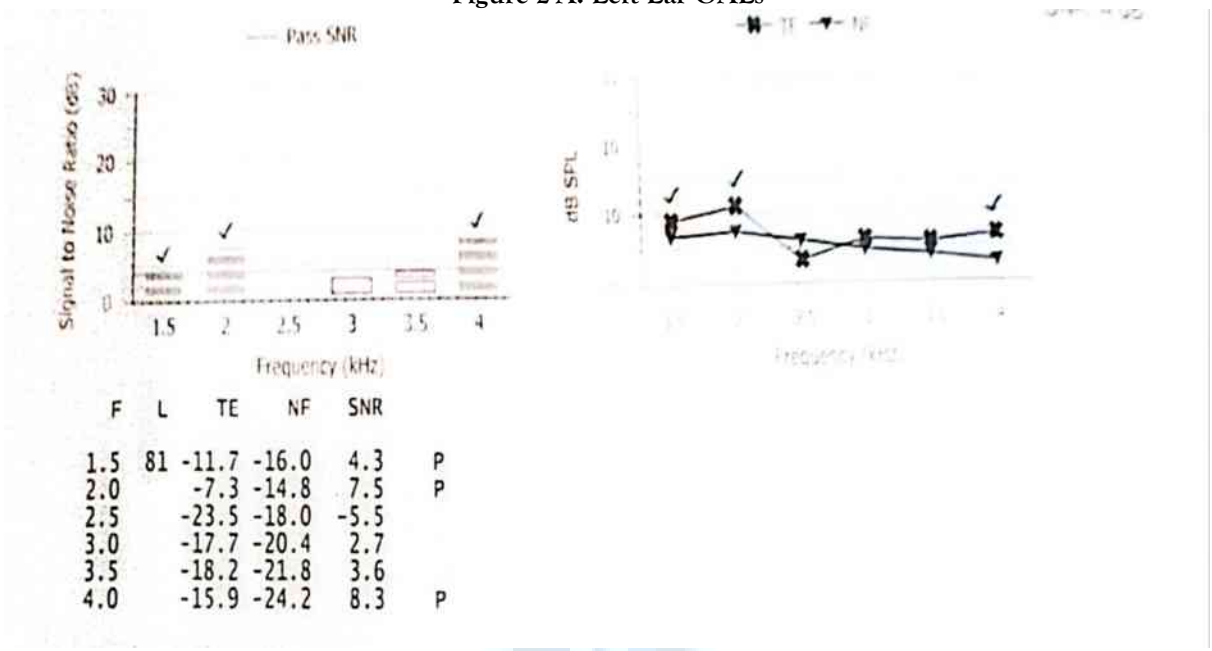
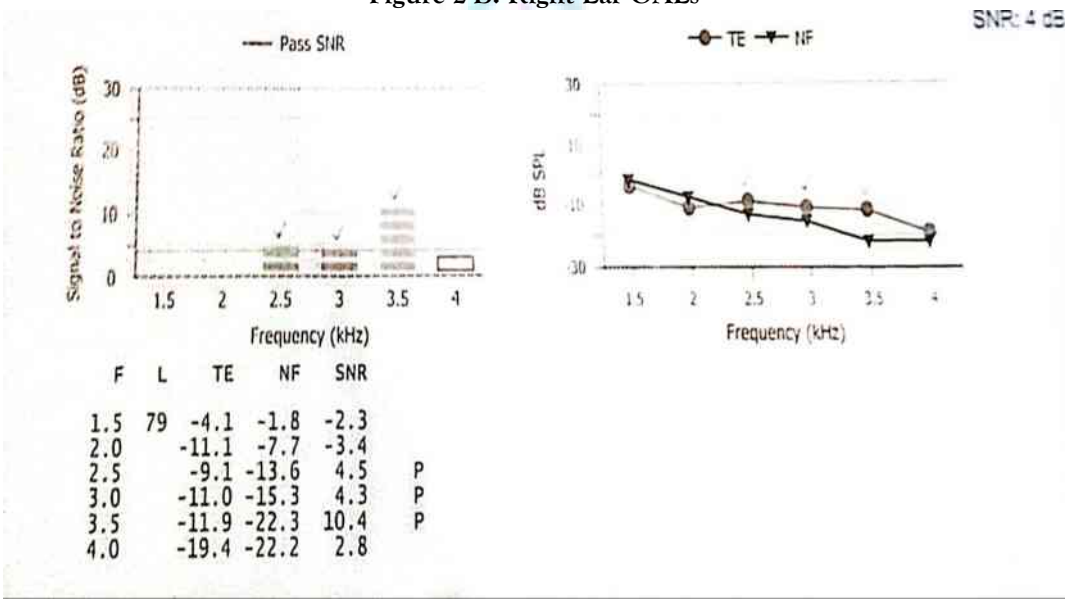


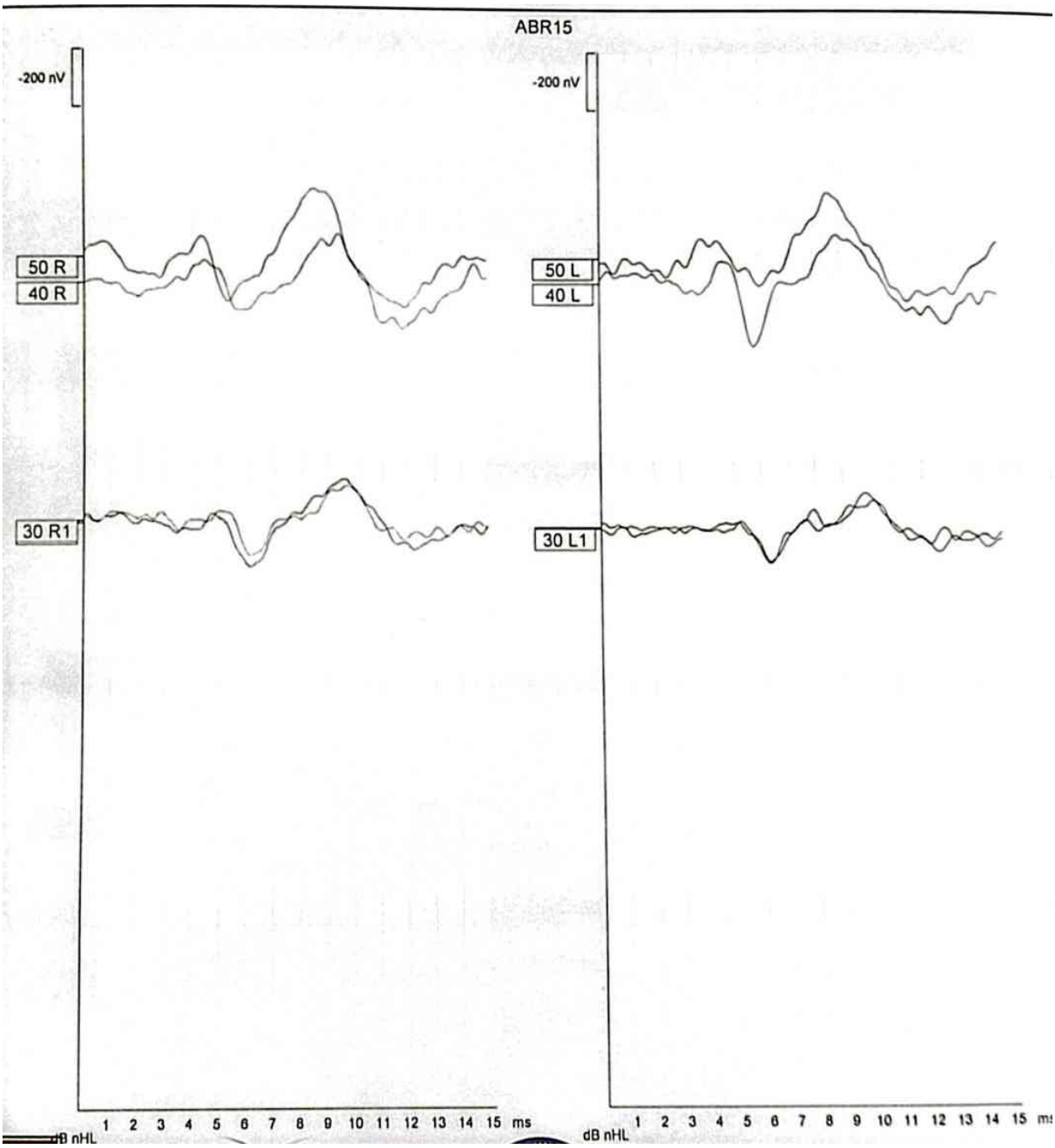
Figure 2-B: Right Ear OAEs



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Figure 3: Auditory Brainstem Response wave morphology

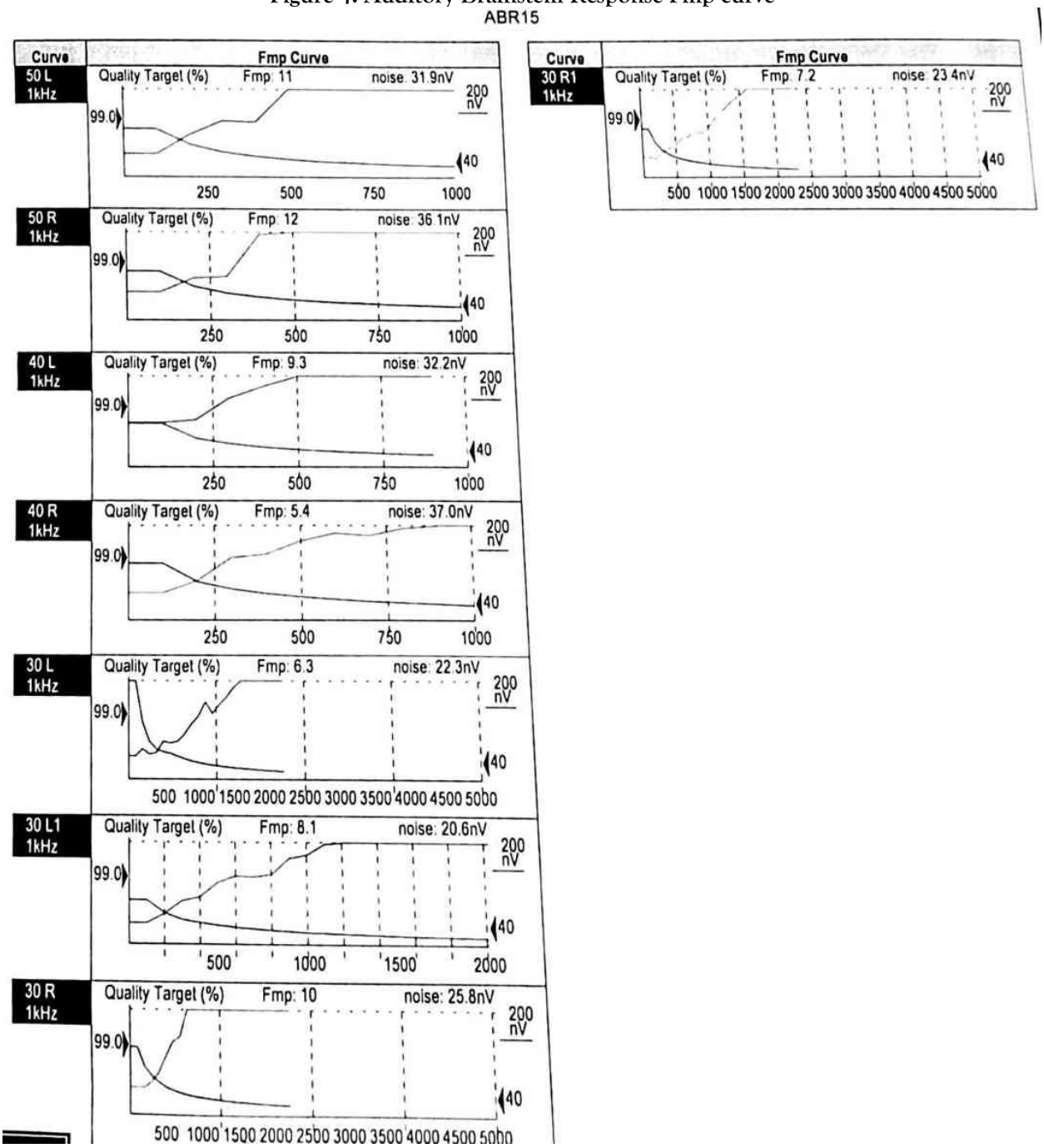


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Figure 4: Auditory Brainstem Response Fmp curve



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Figure 5: Auditory Brainstem Response Audiometry Recorded parameter

ABR15

Curve	Measured Rejected	Gain	Masking	Wave Repro	Stimuli/ sec	Display		Polarity
						Low Pass	High Pass	
50 L HPF 100 Hz 12/oct LPF 3000 Hz ABR3A	1000 0%	±40µV	Off	92 %	44.1	1500Hz	50Hz	CE-Chirp® Alt.
50 R HPF 100 Hz 12/oct LPF 3000 Hz ABR3A	1000 0%	±40µV	Off	93 %	44.1	1500Hz	50Hz	CE-Chirp® Alt.
40 L HPF 100 Hz 12/oct LPF 3000 Hz ABR3A	900 0%	±40µV	Off	96 %	44.1	1500Hz	50Hz	CE-Chirp® Alt.
40 R HPF 100 Hz 12/oct LPF 3000 Hz ABR3A	1000 0%	±40µV	Off	92 %	44.1	1500Hz	50Hz	CE-Chirp® Alt.
30 L HPF 100 Hz 12/oct LPF 3000 Hz ABR3A	2200 0%	±40µV	Off	87 %	44.1	1500Hz	50Hz	CE-Chirp® Alt.
30 L1 HPF 100 Hz 12/oct LPF 3000 Hz ABR3A	2000 0%	±40µV	Off	84 %	44.1	1500Hz	50Hz	CE-Chirp® Alt.
30 R HPF 100 Hz 12/oct LPF 3000 Hz ABR3A	2200 0%	±40µV	Off	95 %	44.1	1500Hz	50Hz	CE-Chirp® Alt.
30 R1 HPF 100 Hz 12/oct LPF 3000 Hz ABR3A	2300 0%	±40µV	Off	93 %	44.1	1500Hz	50Hz	CE-Chirp® Alt.

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Table2: Summary of ABR parameters used

Electrode location	Positive: High forehead Negative :Ipsilateral mastoid Common: Lower forehead
Stimulus type	CE- Chirp
Stimulus polarity	Alternating polarity
Stimulus timing	100µs
Stimulus rate	44.1/s
Amplifier reject levels	±3 to ±10µV peak-to-peak.
Amplifier filters	Low pass filter:1500Hz High pass filter:50Hz
Window length	15ms
Number of sweeps averaged per replication	4000
Display	Wave V up

Discussion:

In the present case, comprehensive objective audiological testing - otoacoustic emissions, tympanometry, and auditory brainstem response - demonstrated normal peripheral and retrocochlear auditory function bilaterally in a 2.4-year-old girl with genetically confirmed DDX3X syndrome. A sound produced by the cochlea is known as an otoacoustic emission (OAE), the generation of which serves as a basic screening tool for hearing loss (15); OAEs in our patient were bilaterally pass, indicating normal outer hair cell function. The auditory brainstem response primarily evaluates the integrity of the auditory nerve and brainstem pathway and is widely used to screen newborns for auditory pathway deficits, identify retrocochlear pathology, and estimate frequency-specific auditory sensitivity (16); in our patient, Wave V was traced at 30 dBnHL bilaterally with a clear response, consistent with normal hearing threshold levels. Tympanometry, an additional diagnostic method for middle ear pathology (17), showed a Type A tympanogram in both ears, indicating normal middle ear function. This finding is consistent with the wider literature, in which auditory abnormalities are a recognized but non-universal feature of DDX3X syndrome, with hearing loss (sensorineural or conductive) reported in only around 8% of affected individuals (13 of 160) across pooled multi-cohort data. Current expert consensus, most notably Levy et al.'s clinical care recommendations, holds that all individuals with DDX3X syndrome should undergo baseline hearing screening, with follow-up audiology assessment reserved for those with recurrent otitis media or a suspected change in hearing acuity- a recommendation that our normal baseline findings, and the surveillance plan we have put in place, directly support. Patient-registry data add further texture to this picture: Simons Searchlight data on males with DDX3X-related disorder describe conductive hearing loss or other ear/hearing defects, and recurrent ear infections in 44% (11/25) of surveyed males (10), suggesting that longitudinal otologic monitoring, rather than a single normal test, is what should ultimately determine whether a child is free of auditory risk. Our patient's Type A tympanograms and clear ABR responses establish a reassuring baseline, but do not preclude the later emergence of conductive pathology from recurrent otitis media, which is common in early childhood generally and may be compounded by hypotonia and craniofacial differences reported in DDX3X syndrome (4). This reinforces our recommendation for structured six-monthly to yearly audiological follow-up rather than a single-visit clearance.

Recent genotype-phenotype work has begun to explain why the auditory and broader somatic phenotype in DDX3X syndrome is so variable. A

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2025 multicenter study of 52 females and 7 males used in silico protein-folding modelling to show that the degree of DDX3X monomer misfolding correlates quantitatively with phenotypic severity, and that variant type and location predict specific phenotypic domains including growth, epilepsy, and brain and motor findings (8). Although auditory outcomes were not separately modelled in that cohort, this variant-severity framework offers a plausible explanation for why some patients, such as the one described here, present with an entirely unaffected peripheral and central auditory pathway despite a confirmed pathogenic DDX3X variant, while others in the literature manifest overt hearing loss. Future case reports and cohort studies would benefit from routinely reporting the specific DDX3X variant alongside audiological findings, so that variant-level genotype-audiophenotype correlations, analogous to those now emerging for growth and neurological features, can eventually be delineated.

DDX3X-related disorder (DDX3X-RD) remains an uncommon monogenic condition. In one cohort, intellectual disability was the most prevalent feature, present in 28 of 31 individuals (18), and a detailed neurodevelopmental evaluation is recommended, since behavioral problems in children with DDX3X syndrome encompass anger, poor impulse control, self-injurious behavior, ADHD, and ASD (19). Speech-language delay was similarly frequent in our patient, consistent with reports that a majority of children with DDX3X syndrome have significant expressive and receptive language impairment, with roughly half of affected girls remaining nonverbal beyond five years of age (7). Normal audiological findings in our patient are reassuring in this context, as they allow the multidisciplinary team to attribute the observed speech delay to central neurodevelopmental factors rather than a peripheral hearing deficit, and permit speech-language intervention to proceed without the confound of unaddressed hearing loss - an important distinction, since hearing loss of any degree in early childhood is independently associated with worse speech, language, educational, cognitive, and social outcomes (11). A thorough speech-language evaluation, and additional periodic hearing examinations given the range of conditions that can cause hearing loss later in life, remain part of our ongoing recommendations for this child.

The 2026 description of a recognizable DDX3X facial gestalt - thin upper lip, flat philtrum, broad nasal bridge, upslanting palpebral fissures, long uniform eyebrows, tapered chin, short neck, and proximal thumb placement (4) - is also relevant to audiologists, since craniofacial dysmorphism is a well-established risk factor for conductive hearing loss via Eustachian tube dysfunction, and, less commonly, for syndromic sensorineural loss. Clinicians evaluating children with these emerging facial features, even before a genetic diagnosis is confirmed, may benefit from a lower threshold for baseline and serial audiological assessment. The 2025 literature also increasingly points to an inflammatory cytokine signature and other emerging biological findings in DDX3X syndrome, raising early prospects for future disease-modifying or precision therapeutics, although these remain at a preliminary, non-clinical stage (9). Work distinguishing the DDX3X behavioral phenotype from idiopathic autism spectrum disorder - particularly the relatively preserved social engagement described in some patients (9) - reinforces the importance of the detailed, multidisciplinary neurodevelopmental evaluation that was already underway for our patient through other specialties on the team.

This case adds several elements not previously reported together for DDX3X syndrome: it is the first documented case from Pakistan, and one of very few published reports internationally to include a complete objective audiological test battery - otoscopy, tympanometry, otoacoustic emissions, and auditory brainstem response - performed under sedation in a single patient with genetically confirmed DDX3X syndrome.

Conclusion:

Our study showed that child with DDX3X Syndrome had normal hearing threshold levels. We advised 6 monthly and yearly follow to monitor hearing threshold for the next 2 years. Referral to speech pathology as part of the multi-disciplinary team was also offered. Detailed

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behavioral and developmental assessment of the child was already undergoing in other specialties of the team.

Abbreviations:

NICU: Neonatal Intensive Care Unit
ERA: EvokedResponse Audiometry
CR: Clear Response
OAE: Otoacoustic Emissions
ABR: Auditory Brainstem Response
ASD: Autism Spectrum Disorder
ADHD: Attention Deficient Hyperactive Disorder

Conflict of Interest:

This article has no conflict of interest.

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