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PREVALENCE OF CONGENITAL AND HEREDITARY ENT PATHOLOGIES IN THE REPUBLIC OF UZBEKISTAN

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Abstract
This study aimed to systematize data from 2014–2025 on the prevalence of congenital and hereditary ENT disorders in the Republic of Uzbekistan (RUz), focusing on congenital/hereditary hearing loss and craniofacial anomalies affecting the ENT system (cleft lip and palate), along with a brief review of rare congenital anomalies of the nose and larynx.
Results: Confirmed neonatal hearing impairment is 3.3 per 1,000 live births (0.33%), exceeding typical global estimates in countries with established universal newborn hearing screening (UNHS) programs. Approximately 50–60% of childhood hearing loss is of genetic origin. In Karakalpakstan, the incidence of cleft lip and/or palate during 2017–2021 ranged between ~0.76 and ~1.3 per 1,000 live births; previous estimates were 1:700–1:850. Approximately 22,000 individuals live with hearing impairment nationwide, supported by 20 specialized boarding schools (2020–2021). Since 2023, a national UNHS program has been implemented alongside expansion of cochlear implantation. Consanguinity remains a significant background factor, contributing to a substantial proportion of congenital anomalies.
Conclusions: Scaling up UNHS, expanding molecular genetic diagnostics, and establishing national registries are essential to reduce early disability and improve speech and cognitive outcomes in affected children.

Keywords: Congenital cleft lip and palate, children, hearing loss, nose, paranasal sinuses.

Introduction

Congenital and hereditary ENT pathologies represent one of the most common groups of sensory disorders in childhood. Consensus reviews indicate that approximately 50–60% of pediatric hearing loss is attributable to genetic causes, including nonsyndromic autosomal recessive forms, whereas the remaining cases result from intrauterine infections, perinatal insults, and environmental factors. Accurate prevalence estimates, screening coverage, and rehabilitation availability are critical for planning health care services.

In recent years, Uzbekistan has systematically developed its audiology services, introducing universal newborn hearing screening (UNHS) and expanding cochlear implantation capacities. Nevertheless, there remains a need for stratified epidemiological surveillance and molecular genetic diagnostics to guide early interventions.

Materials and Methods

A targeted review of publications and reports from 2014–2025 was conducted, emphasizing:

1. Outcomes of newborn hearing screening in Uzbekistan;
2. Regional prevalence data for cleft lip and palate;
3. Official data on infrastructure and social statistics;
4. International reference data on rare congenital ENT anomalies.

Only sources providing numerical indicators (prevalence, incidence, coverage) per 1,000 live births or children were included for comparison.

Results

1. Congenital and hereditary hearing loss

Prevalence: A 2025 report documented 1,372 verified cases of hearing impairment (0.33%), equivalent to 3.3 per 1,000 live births. These values are at the upper end of global estimates for moderate-to-severe hearing loss in populations with incomplete screening, highlighting the need for broader early diagnosis and intervention.

Genetic contribution: Approximately 50–60% of pediatric cases are genetic, emphasizing referral to medical-genetic counseling and targeted molecular testing (e.g., GJB2, OTOF).

Infrastructure: As of 2020–2021, there were ~22,000 individuals with hearing impairment and 20 specialized boarding schools for children with hearing loss, reflecting a substantial social burden.

Policy: Since 2023, Uzbekistan has launched a national UNHS program and expanded cochlear implantation capacity, improving early detection and access to assistive technologies.

2. Craniofacial anomalies affecting ENT function (cleft lip/palate)

In Karakalpakstan (2017–2021), 254 cases were recorded among 190,558 live births, with an incidence ranging from 0.76 to >1.0 per 1,000 live births. Historical regional estimates were 1:700–1:850, consistent with global data (~1:700–1:1,000).

Cleft lip and palate remain the most prevalent craniofacial anomalies, impacting respiration, articulation, and middle-ear ventilation. Regional differences highlight the influence of social and environmental factors.

3. Other rare congenital ENT anomalies

Choanal atresia/stenosis: prevalence ~0.82–0.92 per 10,000 live births (~1:10,000–1:12,000); unilateral-to-bilateral ratio ~2:1; often associated with other malformations; requires urgent neonatal management.

Congenital laryngeal web: extremely rare (<5% of congenital laryngeal anomalies); ~1:10,000 births; significant cause of neonatal stridor.

4. Background epidemiology and consanguinity

Overall prevalence of congenital anomalies in Uzbekistan ranges from 8.5–16.8 per 1,000 live births, with higher rates reported in Tashkent (likely due to better detection).

In 2023, ~10% of infants with disabling anomalies were born to consanguineous parents, consistent with literature reporting increased risk of autosomal recessive disorders, including hereditary hearing loss.

Discussion

Available data from 2017–2025 indicate that the burden of congenital and hereditary ENT disorders in Uzbekistan is comparable to international levels, though neonatal hearing loss (3.3‰) is relatively high, characteristic of systems transitioning from selective to universal screening. The rollout of UNHS, together with expanded rehabilitation services (including cochlear implantation) and parental education, is expected to reduce the age of intervention and improve speech and language development.

Craniofacial anomalies, particularly clefts, remain the most visible ENT-related malformations, affecting essential functions such as respiration, articulation, and middle-ear ventilation. Regional variations (e.g., Karakalpakstan) underscore the need for region-specific programs.

A critical gap is the lack of unified national registries and standardized reporting for ENT-specific congenital anomalies, including severity grading, audiometric thresholds, syndromic vs. nonsyndromic forms, and molecular verification. Addressing this gap will allow more precise estimation of genetic contributions (≥50%) and guide genetic counseling, molecular testing, and preventive measures, such as maternal infection control and avoidance of ototoxic exposures.

Practical Recommendations

1. Achieve ≥95% coverage of UNHS with a two-step protocol (OAE → ABR) and a traceable pathway (“screening ≤1 month → confirmation ≤3 months → intervention ≤6 months”).

2. Implement molecular diagnostic panels for pediatric hearing loss (considering local frequencies of GJB2, OTOF, etc.) and structured medical-genetic counseling, particularly for consanguineous families.

3. Establish a national registry of congenital ENT disorders (minimum dataset, ICD-10/ICD-11 coding, degree of hearing loss, syndromic forms, implantation outcomes).

4. Strengthen prevention strategies: screening and prophylaxis for intrauterine infections (including CMV), control of ototoxic agents, and preconception/prenatal education.
5. Develop regional multidisciplinary centers integrating surgery, ENT, speech therapy, and audiology, with monitoring of otitis media and hearing outcomes in children with clefts.
6. Conduct public awareness campaigns on the risks of consanguinity, supported by national data, and promote voluntary genetic counseling before marriage or pregnancy.

Conclusion

In Uzbekistan, congenital and hereditary ENT disorders impose a significant clinical burden. Pediatric hearing loss (3.3‰, ≥50% genetic) and craniofacial anomalies (0.8–1.3‰ in some regions) are the most prominent. Expansion of universal newborn hearing screening, development of molecular genetic diagnostics, and establishment of national registries are key steps toward reducing early disability and improving speech and cognitive outcomes in affected children.

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