

Congenital Anomalies of The Ear in Newborns from Lviv Region (west Ukraine) for 2006-2018 YY.

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Corresponding author: Kitsera Nataliya, Institute of Hereditary Pathology of National Academy Medical Scientist of Ukraine, Lviv, Ukraine.;E-mail- nkitsera@gmail.com**Received Date:** May 29, 2020; **Accepted Date:** June 02, 2020; **Published Date:** June 12, 2020.**Citation:** Kitsera N. (2020) Congenital anomalies of the ear in newborns from Lviv region (west Ukraine) for 2006-2018 yy. *Journal of Clinical Otorhinolaryngology*, 2(1): Doi: [10.31579/2692-9562/006](https://doi.org/10.31579/2692-9562/006)**Copyright:** © 2020. : Kitsera Nataliya, This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.**Abstract****Objectives:** The ear's congenital anomalies usually occur in the outer, middle or inner ear separately or in combination. We had determined the congenital ear's anomalies (CEA) in newborns (2006-2018 yy) in West Ukraine, Lviv region.**Methods:** 14 newborns who were diagnosed according to the reporting form by the maternity hospitals with CEA were analyzed over a period of 13 years.**Results:** During the study period (2006–2018), there were 366 147 births reported, and 8634 newborns with birth defects, 14 were with CEA, an average incidence of 0.4 per 10 000 births. Our study included 10 boys and 4 girls with CEA. Male-to-female ratio was 2.5:1. The number of newborns with ear pathology was 1.8 times higher in the city than in the village. The most common birth defects of the ear were Q16.1 congenital absence, atresia and stricture of auditory canal (external) – 4 (28.6%) and Q16.9 CEA causing impairment of hearing, unspecified – 4 (28.6%), especially the most common of right side. Microtia (Q17.2) was met very rarely -1 (7.1%) and other CEA Q16.9-1 (7.1%). During this period there were only 2 cases of anotia and 1 case of microtia. There was only 1 (7.1%) case of bilateral CEA in boy from town. Just one girl had CEA was associated with birth defect of musculoskeletal system.**Conclusion:** This is necessary to establish the correct diagnosis in time, especially for mind of the family doctors, genetic counseling for relative's family for has benefited earlier prophylaxis.**Advances in Knowledge:** The ear's congenital anomalies usually occur in the outer, middle or inner ear separately or in combination. So congenital microtia met as an isolated defect or is one of many genetic syndromes.**Application to Patient Care:** The study is also important as it may help to raise the awareness of surgical pediatric intervention and to emphasize the loss of babies with congenital abnormalities.**Keywords:** congenital anomalies: congenital ear's anomalies**Introduction**

The baby, at the moment of the birth often diagnose with microtia or anotia compared to other ear defects [1-4]. Microtia is one of the rarely congenital anomalies, which has various from complete absence (anotia) or mild defects of the external meatus. According to ICD-10-CM congenital malformations of ear causing impairment of hearing posted in section Q16.0-Q17.9 [5].

The storages of outer ear consist of three parts: the external ear (which include ear pinna and auricle), the ear canal (which respond external acoustic) and the outer layer of the tympanic membrane (which called "eardrum"). Mesenchyme of the first and second pharyngeal arches provide outer ear development. At list part, it controlled by genes, which determine these pharyngeals arch identity. In generous, the pathogenesis of this anomaly still not understood at all. In the most part of cases, whole deviations from the normal development of the ear structure, happen by first half of pregnancy. Sometimes, congenital anomalies of the ear combine with skull's or face anomalies, or another birth defects [6,7]. If child's congenital ear anomaly go together with another birth blemish, it may be signal for existance of the genetic syndrome [8,9].

When abnormality manifests only in cosmetic defect, it doesn't have serious effect for child's health, because it can be correct by plastic surgery. However, when cosmetic defect accompanied by hearing loss,

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this case become more difficult as child's socialization start to be harder [8,10,11].

Congenital birth defects of ear are sometimes can de part of genetic syndrome or happen in isolated cases. It's frequency composes 0.3–0.6 per 10 000 live births [12,13]. The most common form is unilateral microtia, especially right-sided microtia [14]. EUROCAT submits cases and prevalence (per 10,000 births) for all full member registries from 2011 to 2017 of anotia - 0.39 [15]. Prevalence in Ukraine of this pathology was 0.38 per 10 000 newborns in 2011-2017 yy [15]. At the same time according to EUROCAT frequency of all birth defects in Ukraine was 250.76 per 10 000 newborns during this period.

Mother from Latin American, Chinese, American Indian and Filipins are the biggest carriers of the genes, which induce birth of children with this pathology [11,16,17]. If we look at the statistics, we can see that female with that anomaly dominate by male, especially in isolated forms [16]. But risk of associated congenital anomalies with microtia was highest in female [16]. It's development is influenced by the following reasons [18,19]. One of the basic reasons of the outer ear anomaly are: the influence of hereditary or genetic factors or the environment, for example using alcohol by woman while she is pregnancy or taking drug[20].

Microtia is often accompanied by multiple congenital anomalies. The most common associations are anomalies of the extremities, cleft lip

and/or palate, birth defects of cardiovascular system, vertebral defects [21].

The most common syndromes associated with microtia are Townes–Brock syndrome, Goldenhar syndrome (oculoauriculovertebral spectrum), Treacher Collins and branchio-oto-renal syndrome [6, 22, 23].

The majority of congenital anomalies of ear associated with another malformations: oculoauriculovertebral syndrome (Goldenhar syndrome, characterized by incomplete development of the ear, nose, soft palate, lip and mandible caused by *HMX1* mutations), Treacher Collins (associated with deformities of the ears, eyes, cheekbones, and chin, caused by mutations in *TCOF1*, *POLIRC*, *POLIRD* genes), Branchio-oto-renal or Melnick-Fraser syndrome (kidneys, ears and neck are affected by mutations in *SIX5*, *EYAI*), Mandibulofacial dysostosis with microcephaly (developmental delay and abnormalities of the head and face, small and abnormally-shaped ears for *EFTUD2* mutations), CHARGE syndrome (affects many areas of the body - coloboma, heart defects, atresia choanae, growth retardation, genital and ear abnormalities associated with mutations in *CHD7* gene), Lacrimo-auriculo-dental-digital or LADD syndrome (aplastic/hypoplastic lacrimal and salivary glands and ducts, cup-shaped ears, hearing loss, hypodontia enamel hypoplasia, distal limb segments anomalies caused by *FGFR2*, *FGFR3*, *FGF10* genes), Townes–Brocks Syndrome (imperforate anus, hands and ears malformations and kidney abnormalities, caused by *SALL1* mutations), Kabuki syndrome (affects multiple parts of the body with varying symptoms including ear deformity microtia associated with mutations in *MLL2*, *KDM6A* genes) [14, 21, 22, 24].

The aim of our research was to determine the congenital ear’s anomalies in newborns around 2006-2018 yy in West UA, that’s why the objectives of explores were from Lviv region.

Materials and methods

We collected a retrospective cohort, exploited the reports, which assembled from maternity hospitals dictates gathering in Lviv region over a period of 13 years. All maternity hospitals granted Ethical Permission for this study. Data were obtained in accordance with the free and conscious wish of the mother of child with birth defects on the basis of her informed consent. An obstetrician in a maternity hospital who is responsible for the delivery of such a pregnant woman and a neonatologist

examining the baby informed her of the peculiarities of collecting and using data about the child with birth defects before making informed consent. Informed consent was made at the maternity hospital before the baby is born. This report does not contain any personal information that could lead to the identification of the patient.

We identified 14 patients, who have ear’s anomalies from their birth, with or without associated malformation. Infant sex was collected as male or female. Infant sex was collected as male or female. Our epidemiological explorations about the congenital anomalies of the ear, were pithy, especially external ear malformations anotia and microtia. The purpose of our research was to reveal nuances of the relationships between sex, type of microtia, and birth place and associated malformation as seen in Lviv region.

Results

In accordance to the Statistical Office on January 2018, [25] population was 2 511 238 persons, including 1 190 549 men (47.4%) in Lviv region (West Ukraine). Every year in the Lviv region were born from 30 270 to 26 452 children. Congenital malformations were diagnosed more than five hundred newborns each year, and sometimes more than 900 children [Table 1]. The procent of whole birth defects had been from 1.8 % 2008 year to 3.5% in 2006-2007 years in Lviv region, which is the same to the general population fact in UA [26].

We took newborns from Lviv town and 20 districts of Lviv region (2006-2018 yy) respectively to the medical records from maternity hospitals. Congenital anomalies of the ear was identified in 14 newborns [Table 1]. This pathology can be seen immediately after the baby is born.

A total of 366 147 births over 13 consecutive years from 2006 to 2018 were included in the study and 8634 newborns had congenital anomaly, an average incidence of 235.81 per 10 000 births. The prevalence estimates (per 10,000 live births) were 0.4 (14 cases). The lowest frequency of congenital anomalies of the ear was observed 0.3 per 10000 newborns in 2012-2013 years. Very high frequency of this pathology – 2.2 per 10000 newborns was diagnosed in 2007 year. In some years [Table 1], no cases of the specified pathology were detected, including two consecutive years -2014-2015 yy. Every year in Lviv region, there are few children, which born with this pathology [Table 1].

	2006	2007	2008	2009	2010	2011	2012	2013	2014	2015	2016	2017	2018	2006 - 2018
Number of newborns	27272	27454	29007	30079	28651	28904	30220	29542	30270	27909	27134	26452	23253	366147
All BD	958	948	525	641	643	656	711	695	664	633	575	495	490	8634
%	3.5	3.5	1.8	2.1	2.2	2.3	2.4	2.4	2.2	2.3	2.1	1.9	2.1	2.4
congenital anomalies of the ear	1	6	-	2	-	1	1	1	-	-	1	-	1	14
Frequency of congenital anomalies of the ear 1:10 000	0.4	2.2	-	0.7	-	0.4	0.3	0.3	-	-	0.4	-	0.4	0.4

Table 1. Frequency of congenital anomalies of the ear in newborns from Lviv region (West Ukraine) by year, 2006 to 2018

The largest number of cases – 6, was registered in 2007 year. We were diagnosed with 1 case of congenital anomalies of the ear in newborns in 2011-2013 yy.

In the group of our researching were 10 boys and 4 girls with congenital anomalies of the ear (5 from village and 9 newborns from town). There

wasn't any case of multiple pregnancy from newborns with this pathology. This group was characterized by a male predominance of 71.4% - 10 cases [Table 2] and diagnosed only in 4 (28.8%) girls. Male-to-female ratio was 2.5:1.

Q	Diagnosis	n	%	Man	Female	From town	From village
Q16.0	Congenital absence of (ear) auricle	2	14.3	1	1	2	-
Q16.1	Congenital absence, atresia and stricture of auditory canal (external)	4	28.6	3	1	3	1
Q16.4	Other congenital malformations of middle ear	1	7.1	1	-	-	1
Q16.9	Congenital malformation of ear causing impairment of hearing, unspecified	4	28.6	3	1	3	1
Q17.2	Microtia	1	7.1	1	-	-	1
Q17.9	Congenital malformation of ear, unspecified	2	14.3	1	1	1	1
All		14	100	10	4	9	5

Table 2. Spectrum of congenital anomalies of the ear in newborns from Lviv region (West Ukraine) by year, 2006 to 2018.

The most common birth defects of the ear [Table 2] were Q16.1 congenital absence, atresia and stricture of auditory canal (external) – 4 (28.6%) and Q16.9 congenital malformation of ear causing impairment of hearing, unspecified – 4 (28.6%) in newborns from Lviv region (West Ukraine), specially the most common of right side. Microtia (Q17.2) was met very rarely -1 (7.1%) and other congenital malformations of middle ear Q16.9-1 (7.1%). Two cases of congenital absence, atresia and stricture of auditory canal (external) – Q16.1 were established in 2007. During this period there were only 2 cases of anotia and 1 case of microtia.

A retrospective chart review was performed for all cases of congenital anomalies of the ear in newborns, according to the reporting form submitted by the maternity hospitals from Lviv region (West Ukraine) over the period 2006-2018 yy showed that is significant associations were observed between geographic areas. This pathology was more often diagnosed by patients whose parents resided in the city (9 cases -64.3%) as opposed to rural citizens (5 cases-35.7%).

The Table 3 presents counts specifics of localisation of congenital anomalies of the ear among the newborns, 92.9% had unilateral defects.

Congenital anomalies of the ear	n	%	Another birth defect	
Unilateral, left	3	21.4		
Unilateral, right	4	28.6		
Unilateral, not specified	6	42.9	1	Congenital talipes equinovarus
Unilateral—total	13	92.9		
Bilateral	1	7.1		
Total	14	100	1	

Table 3. Specifics of localization of congenital anomalies of the ear and other birth defect in newborns from Lviv region (West Ukraine) by year, 2006 to 2018.

The right ear is affected in 28.6% of individuals than left-side defect-21.4%. There was only 1 (7.1%) case of bilateral congenital anomalies of ear in boy from town. In one girl with unilateral not specified congenital anomalies of the ear was associated with other birth defect of musculoskeletal system - congenital talipes equinovarus. Our results show a slightly lower proportion of congenital anomalies of the ear with co-occurring defects compared to isolate this pathology.

Discussion

This research provides recent population-based frequency estimates for ear anomalies among 366 147 newborns from Lviv and 20 districts of Lviv region (West Ukraine). Decreasing trend of the birth prevalence of anotia and microtia is observed in Lviv region (West Ukraine). In addition, our analysis provides new data about spectrum of birth defects of ear and association with other congenital anomalies in our region.

Our results show a slightly higher proportion of anotia/ microtia as isolated anomaly. This data is the same from what was observed by Deng K. et al. [12] who found a higher prevalence of isolated anotia/microtia.

We diagnosed a higher proportion of unilateral congenital anomalies of the ear in newborns from Lviv region compared to bilateral cases. However, among the dictates of other authors¹⁸, we observe that the frequency of bilateral ear defects is higher -21% (Stallings EB et al.) than in our study -7.1%.

Around reports of other authors we can observe that our results are similar to previous studies of birth defects of ear with respect to the prevalence infant sex [12, 17].

If we simile the high incidence of congenital anomalies of the ear in newborns among China and Spanish, in our sample residents of Lviv region frequency is lower. Perhaps, the discrepancy of this pathology is related to the ethnic features of each population [7,8,11,12,17]. Canfield M. A. et al. [7] notes that the frequency birth defects of ear is 2.86 per 10 000 live births (Texas, USA) but frequency of this pathology in Lviv region (West Ukraine) in 2006-2018 is 0.4 on 10 000 newborns. This frequency is the same in Lviv region as in other regions of Ukraine - 0.42 and in Belgium -0.38, Poland -0.33 but higher than in Netherlands -0.1, Spain -0.12, Portugal-0.13, Ireland -0.15, Croatia-0.19, and lower in such countries as Switzerland -0.68, Hungary -0.69, France (Paris) -1.07[15].

Congenital ear abnormalities are a group of diseases of varying severity, especially when it is related to hearing, and many require a multidisciplinary approach for better diagnosis and accurate treatment.

Since the spectrum of congenital ear abnormalities is different and heterogeneous various studies predominate in variety birth defects [6,12,19,21]. In our study prevalence congenital absence, atresia and stricture of auditory canal (external) and congenital malformation of ear causing impairment of hearing, unspecified. Most often diagnosed among all the defects - congenital absence, atresia and stricture of auditory canal (external) - Q16.1 4 cases and Congenital malformation of ear causing impairment of hearing, unspecified - Q16.9 - 4 cases.

In addition, patient's doctor must pay attention for impairment of the hearing, cause it is momentous for timely diagnostic. Also, it preclude complications of disease. That's why ill child resave the opportunity treatment. It is momentous for relatives of the patients, which must go to genetic counseling with destination of prophylaxis of this pathology among subsequent generations and sibling.

Our research were the largest longitudinal explores of microtia allowing for demographic and comorbidity commentary.

Conclusion:

We observed 14 cases among newborns which had congenital anomalies of the ear. They were diagnosed till 13 years (2006-2018) from Lviv region (West Ukraine) and frequency was 0.4 on 10 000. The frequency coincides with the data of many European countries ethnically similar population, for example in particular with Poland bordering on the Lviv region. But spectrum of congenital anomalies of the ear is very different. The number of newborns with ear pathology was 1.8 times higher in the city than in the village. But spectrum of congenital anomalies of the ear is very different. Just one patient (7.1%), who had congenital ear's anomalies, was associated with birth defect of musculoskeletal system. This is necessary to establish the correct diagnosis in time, especially for mind of the family doctors.

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