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Inner ear malformations in congenital deafness are not associated with increased risk of breech presentation

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Running title: inner ear and breech presentation

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## **Abstract**

As there is speculation in the literature that function of an immature vestibular system may be associated with cephalic presentation at delivery, our aim was to determine whether syndromes with congenital inner ear malformations were accompanied by a higher frequency of breech presentation/malpresentations than in general population (2-3%). **METHOD:** A review was undertaken of free MEDLINE (1936-2016) to determine frequency of breech presentation and transverse lie in cases with congenital deafness (Michel aplasia, Wildervanck syndrome, Mondini-Alexander dysplasia, Waardenburg syndrome, CHARGE syndrome, Large vestibular aqueductal syndrome, Pendred syndrome, Oculo-aurico-vertebral spectrum, Jervel and Lange-Nielsen syndrome, Usher syndrome, Scheibe dysplasia) and vestibular nerve aplasia. **RESULTS:** Identified were a total of 122 cases. The frequency of breech presentation was 1.63%, and of transverse lie 1.63%, in total 3.28% malpresentations. **CONCLUSION:** The results of study suggest that congenital malformations of the vestibular apparatus are not associated with the increased risk of breech presentation at delivery.

**Key words:** congenital deafness; inner ear malformation; breech presentation; vestibular malformation; fetus; gestation.

## **Introduction**

The fetus changes its presentation by active whole-body movements, such as kicking (1). Fetal kicking represents a sudden extension of the trunk and lower limbs, as a result of excitations of the extensor muscles. Vestibular nuclei, which are under constant afferent activity of the vestibular apparatus, excite extensor muscles of the trunk and lower extremities (2). The vestibular labyrinth acquires adult morphology and size during the 5<sup>th</sup> gestational month (3). The myelination of the vestibular nerve begins in the 17<sup>th</sup> gestational week and ends prenatally (4).

Congenital deafness is accompanied by abnormalities in the function of the vestibular apparatus in 80-85% of cases (5,6). Pathological substrates of the congenital deafness are complex malformations of the cochlea and vestibular apparatus.

Our study aimed at determining whether syndromes with congenital inner ear malformations are accompanied by a higher frequency of breech presentation/malpresentations than in general population 2-3% (7) by reviewing existent published scientific data.

## **Material and methods**

Free MEDLINE/PubMed search was undertaken in November 2016. The key words of the stated inner ear malformations, and syndromes according to Omerod's classification (8) are: “Michel aplasia”, “Wildervanck syndrome”, “Mondini-Alexander dysplasia”, “Waardenburg syndrome”, “CHARGE syndrome”, “Large vestibular aqueductal syndrome”, “Pendred syndrome”, “Oculo-aurico-vertebral spectrum”, “Jervel and Lange-Nielsen syndrome”, “Usher syndrome” and “Scheibe dysplasia”. Considering that vestibulocochlear nerve aplasia leads to a

complete deafferentation from the vestibular apparatus, we included this medical entity in the search with key words: “aplasia” “vestibulocochlear nerve” and “vestibular nerve”.

In addition to the term cephalic presentation, the terms “uneventful birth”, “unremarkable birth history”, “normal vaginal delivery”, “uncomplicated delivery”, “normal delivery”, “uneventful perinatal” and “uneventful intranatal period”, were considered as cephalic presentation at birth. We included publications that described case reports or series of case reports with data of fetal presentation in single-fetus pregnancies. Two authors reviewed the findings to reduce the risk of selection bias. First, the two reviewers screened the publications by title and abstract. Publications which fulfilled the inclusion criteria were screened in full text.

## **Results**

Research covered period from 1936 to 2016 year. The literature search resulted in n=5837 citations (after de-duplication). In the first stage n=5579 articles were excluded by review of title and abstract. At the second stage, 258 articles met the eligibility criteria based on title and the corresponding full-text articles were procured for review. Finally, n=91 papers with a total of 122 cases were included in our study. Malformation according to reference numbers were: Michel aplasia (9-11), Wildervanck syndrome (12-26), Mondini-Alexander dysplasia (9,17-25), Waardenburg syndrome type I (26-36), Waardenburg syndrome type II (37-41), Waardenburg syndrome type III (42-46), Waardenburg syndrome type IV (47-54), CHARGE syndrome (55-63), Large vestibular aqueductal syndrome (64-71), Pendred syndrome (72-77), Oculo-aurico-vertebral spectrum (78-85), Jervel and Lange-Nielsen syndrome (86-90), Usher syndrome (91 - 94), Scheibe dysplasia (95), and Pontine tegmental cap dysplasia (96-99).

**Table 1. around here.**

The frequency of breech presentation was 1.63% and of transverse lie 1.63%, giving a total of 3.28% (Table 1.).

Table 2. presents cases derived from the first table in which malformations of the vestibular apparatus were described (utricle, saccule, semicircular canals) based on radiological examinations (Computed Tomography or Magnetic Resonance Imaging). Breech presentation and transverse lie was not reported in any of these cases.

**Table 2. around here.**

Only in five cases, one with Wildervanck (16) and four with Mondini-Alexander dysplasia (25), provided data about the vestibular function. In all five cases presentations were cephalic and in all five cases caloric examination revealed total absence of vestibular response bilaterally (16,25). Damped rotation test elicited per-rotatory nystagmus in the four examined children with Mondini-Alexander dysplasia but the duration and total number of nystagmic beats were decreased compared with healthy children of the same age (16,25).

**Discussion**

This paper is the first to provide a comprehensive overview of existing studies on inner ear malformation in congenital deafness and its association to breech presentation. The obtained frequency of breech presentation in congenital deafness was 1.63%, while in the general population it is 2% -3% (7).

Prior current study, only one other study published data related to congenital deafness and malpresentations (100). This article did not analyze specific syndromes of congenital deafness.

The obtained frequency of breech presentation in this study does not differ from the data reported in the present study. Those findings together, indicate that the abnormal function of the vestibular apparatus was not associated with an increased risk of breech presentation.

In the examined types of congenital deafness there were a high proportion of cases with abnormal function of the vestibular apparatus. In four out of ten subjects with Waardenburg syndrome and hearing impairment the torsion swing test showed a directional preponderance of nystagmus. The caloric test was abnormal in five out of ten patients (101). The frequency of abnormal function of the vestibular apparatus in CHARGE syndrome was 100%; however, a residual response could be registered in 94% (102,103). Large vestibular aqueductal syndrome was also often accompanied by abnormal vestibular function. In another study investigating the association between Jervell and Lange-Nielsen Syndrome (JLNS) genotype and vestibular dysfunction, an abnormal post-rotatory finding was seen in 9/9 examined cases, while 3 of them had a complete lack of vestibular function (104). In 16 examined patients with Usher syndrome Type I there was no nystagmic response on the caloric test, while the results of the rotatory test were abnormal in 11 patients (105).

However given the presence of the residual function of the vestibular apparatus in cases of congenital inner ear malformation, it is possible that the residual function of the vestibular apparatus is sufficient for occupying cephalic presentation in the second half of gestation.

Beside the vestibular apparatus, vestibular nuclei receive input from proprioceptive, visual, tactile, and auditory sources. Based on all these sensory inputs, the vestibular system generates efferent motor activity (2). Characteristic of the vestibular nuclei is spontaneous discharges even in the absence of afferentation by the vestibular apparatus (106). Also, the plasticity of the central nervous system provides the possibility of compensation after the

vestibular apparatus has lost its function (2). All this suggests that even when the function of the vestibular apparatus is absent, the vestibular system can still be functional.

The results of our study suggest that congenital malformations of the vestibular apparatus and its abnormal function do not increase risk of breech presentation. The residual function of the vestibular apparatus could be sufficient for fetus to occupy cephalic presentation. When the function of the vestibular apparatus is absent, the vestibular system can still take part in regulation of fetal presentation because of sensory afferentation from proprioceptive and cutaneous system.



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## **Declaration of Interest**

The authors report no conflicts of interest

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Table 1. Presentation and lie at delivery in cases of vestibulocochlear dysplasias with associated disorders.

Description of malformations	Malformations and associated syndroms	CP No	BP No	TL No
Complete lack of inner ear development	Michel aplasia	3	0	0
	Wildervanck syndrome	5	0	0
Cochlea contains a decreased number of turns and underdevelopment of vestibular structures	Mondini-Alexander dysplasia	12	0	0
	Waardenburg syndrome Type I	13	0	0
	Waardenburg syndrome Type II	5	0	0
	Waardenburg syndrome Type III	6	0	0
	Waardenburg syndrome Type IV	8	0	0
	CHARGE syndome	17	0	2
Large vestibular aqueductal syndrome	Large vestibular aqueductal syndrome	11	1	0
	Pendred syndrome	6	0	0
Malformation restricted to the membranous labyrinth. Normal bony labyrinth	Oculo-auriculo-vertebral spectrum	10	0	0
	Jervel and Lange-Nielsen syndrome	13	0	0
	Usher syndrome Type I	4	0	0
Malformation of the membranous cochlea and saccule	Scheibe dysplasia	1	0	0
Aplasia of the vestibular nerve	Pontine tegmental cap dysplasia	4	1	0
Total cases		118	2	2

CP – Cephalic Presentation, BP – Breech Presentation, TL – Transverse Lie, No – number of cases

Table 2. Types of congenital vestibular malformations in congenital deafness and association with breech presentation.

Case number	Sacculus		Utriculus		Horizontal Semicircular Canal		Anterior Semicircular Canal		Posterior Semicircular Canal		BP	Reference number
	D	A	D	A	D	A	D	A	D	A		
1						+		+		+	No	8
2						+		+		+	No	9
3		+		+		+		+		+	No	10
4						+		+		+	No	16
5					+						No	16
6	+		+		+		+		+		No	16
7					+						No	17
8					+		+		+		No	18
9						+		+		+	No	19
10				+		+		+		+	No	20
11				+		+		+		+	No	21
12				+		+		+		+	No	23
13						+		+		+	No	24
14								+		+	No	24
15								+		+	No	24
16						+		+		+	No	24
17					+					+	No	36
18						+		+		+	No	50
19						+		+		+	No	51
20	+		+		+		+		+		No	81
21	+		+		+						No	82
22	+										No	94

BP – Breech presentation, D – Dysplasia, A – Aplasia.

