EVOLUTION OF KNOWLEDGE ON CRANIOVERTEBRAL ANOMALIES AND ARNOLD–CHIARI MALFORMATION: HISTORICAL, MORPHOLOGICAL, AND DIAGNOSTIC PERSPECTIVES

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Abstract: *The study explores the historical development, classification, and diagnostic evolution of craniovertebral anomalies with a focus on Arnold–Chiari malformation. Over the past three centuries, the understanding of these conditions has progressed from descriptive anatomical observations to detailed radiological and genetic analyses. The work traces the contributions of early anatomists such as R. Columbus, H. Chiari, and J. Arnold, and the refinement of classifications by modern researchers. Special attention is given to the morphogenesis of the craniovertebral junction, the relationship between bone malformations and posterior fossa size, and the pathogenesis of cerebellar tonsil descent. Despite advancements in MRI diagnostics, the etiopathogenesis of Arnold–Chiari malformation remains controversial, with competing theories involving hydrocephalus, traction, and osteoneural disproportion. This review highlights the complex interplay of embryological, anatomical, and radiological factors that continue to shape current diagnostic and clinical approaches.*

Keywords: *craniovertebral junction, Arnold–Chiari malformation, atlas assimilation, basilar impression, platybasia, MRI diagnosis, embryogenesis, posterior cranial fossa.*

Introduction. The history of studying craniovertebral anomalies spans more than three centuries. Initially, the analysis of this pathology was conducted by anatomists and was mainly descriptive in nature. The first description of atlas assimilation is attributed to the Italian anatomist R. Columbus [1]. In 1790, J. Ackermann described in detail the developmental anomaly of the main part of the occipital bone, subsequently named basilar impression by F. Berg and A. Retzius (1855) [1, 2]. Later, in the 19th century, R. Virchow described platybasia, in which

an underdeveloped clivus assumes a horizontal position, thereby causing flattening of the skull base. Bone anomalies in the craniovertebral region are quite often combined with Arnold-Chiari malformation [3, 4]. In recent decades, due to the rapid development and introduction of modern methods of radiation diagnostics into clinical medicine, and advances in the field of genetics and embryology, it has become possible to conduct an in-depth study of craniovertebral junction dysplasia, since this pathology still remains a complex problem that is not fully understood [5]. In a historical essay, J.M. Pearce (2000), citing the article "Contributions to the study of spina bifida, encephalocele, and anencephalus" in the London Journal of Anatomy and Physiology, reports that in 1883, J. Cleland published information on the combination of elongation of the brainstem and descent of the cerebellar tonsils into the spinal canal with hydrocephalus and spina bifida in a newborn. More than 50 years earlier, Parisian professor of anatomy J. Cruveilhier presented a case of a combination of cerebellar and medulla oblongata herniation with myelomeningocele and diastematomyelia in an anatomical atlas. Then, in 1891, Prague professor of pathology H. Chiari characterized three types of malformation. Description of the first type includes the observation of elongation of the cerebellar tonsils and their displacement into the spinal canal, while the fourth ventricle remains unchanged or slightly elongated, in addition, flattening of the medulla oblongata is sometimes observed [6,7] (Fig. 1). The second type is characterized by a downward displacement of the spinal cord, cerebellar vermis, fourth ventricle and lower parts of the brainstem below the level of the foramen magnum, combined with other dysraphisms: aqueduct stenosis and hydrocephalus, underdevelopment of the tentorium cerebelli. These changes are almost always combined with lumbosacral myelomeningocele [2, 8] (Fig. 2). Anomaly of the third type is rare and is manifested by a gross displacement of the cerebellum and medulla oblongata into the spinal canal [9]. In 1894, R. Virchow's student J. Arnold published his observations of changes in the cerebellum and medulla oblongata in newborns with spina bifida. In 1986, H. Chiari supplemented his classification by including a fourth type, which was characterized by severe cerebellar hypoplasia without protrusion of the tonsils

into the foramen magnum [7,8]. Similar cases were described by the Russian neurologist N. N. Solovtsov [10]. In 1907, E. Schwalbe and M. Gredig proposed the eponym "Arnold-Chiari malformation". However, some authors,Taking into account the smaller contribution of J. Arnold to the description of this pathology, they consider it correct to call it the term "Chiari malformation" for all types [9]. According to M.D. Blagodatsky et al. (1995), the frequency of detection of Arnold–Chiari malformation type I in the population is 3.3–8.2 per 10,000 population. According to the study conducted by N.E. Krupina et al. (2002), when analyzing 13,500 magnetic resonance tomograms of the craniovertebral region of patients with a neurological profile, a high incidence of Arnold–Chiari malformation I was noted - in 3.9%. There is an assumption about gender-dependent distribution of craniovertebral anomalies, since the incidence in men and women is not the same, women predominate (the ratio of occurrence in males and females, according to T.A. Akhadov (1993), is 41% and 59%, respectively, according to N.E. Krupina (2002) - 2:3, according to AD Elster, MY Chen (1992) - 1: 2). In Arnold-Chiari malformation type I, according to data from different authors, basilar impression, assimilation of the atlas, and platybasia were most frequently diagnosed [1,3,11]. The classification of bone anomalies of the craniovertebral junction is based on various principles of systematization. AH Menezes divides all pathologies of the craniovertebral junction into congenital and acquired, taking into account the etiology and the zone of damage to the craniovertebral region. He classifies the following as congenital malformations: 1) malformations of the occipital sclerotomes (basilar invagination, hypoplasia of the condyles, etc.); 2) malformations of the atlas (assimilation of the atlas, aplasia of the atlas arches, etc.); 3) malformations of the axis (dysplasia of the odontoid process, etc.). Acquired disorders and developmental anomalies of the CVP are subdivided into pathology of the foramen magnum (the latter includes secondary basilar invagination, as a consequence of Paget's disease, rickets, rheumatoid arthritis, etc., and foraminal stenosis) and atlantoaxial instability (the cause of which can be infections, injuries, tumors, etc.) [12]. The classification by D. Pang et al. (2011) is based on the embryogenesis of the craniovertebral junction

components. All bone malformations are divided into two groups: 1) malformations of the central structures caused by disruption of the formation of the axial components of the occipital sclerotomes, proatlas, and first cervical sclerotome (these include dysgenesis of the odontoid process, "odontoid bone," aplasia/hypoplasia of the odontoid process components, as well as basilar impression, platybasia, etc.); 2) malformations of the surrounding structures caused by impaired development of the lateral components and hypochordal regions of the proatlas and first cervical sclerotome (formation of the third occipital condyle, hypertrophy of the occipital condyle, aplasia and hypoplasia of the anterior arch of the atlas, agenesis of the posterior arch of the atlas, assimilation of the atlas, aplasia of the lateral masses, etc.) [13]. Congenital pathology of the cervicoccipital region has long been considered as anatomical curiosities [14],which was caused by the difficulties of instrumental diagnostic methods. The active and widespread use of computed tomography and magnetic resonance imaging has made it possible to study in detail the most complex anomalies of the brain. However, to date, there is no consensus regarding the radiological criteria for establishing the diagnosis of Arnold–Chiari malformation type I. In 1985, A. O. Aboulezz et al. first proposed MRI criteria for diagnosing Arnold–Chiari malformation: dystopia of the cerebellar tonsils below the level of the foramen magnum by more than 5 mm should be considered pathological, the position of the tonsils between 3 and 5 mm is a borderline condition, and a descent of up to 3 mm is considered normal [6]. In 1986, AJ Barkovich et al., while studying magnetic resonance imaging of the brain of two hundred patients and comparing them with the clinical picture, came to the conclusion that ectopia of the cerebellar tonsils of more than 3 mm can be used to establish the diagnosis of Arnold–Chiari malformation type I. In 1992, AD Elster and MY Chen, and in 2000, J. Meadows noted that the descent of the cerebellar tonsils below the foramen magnum from 5 to 12 mm in 24–31% of patients can be asymptomatic [6]. DJ Mikulisetal. (1992), analyzing the results of magnetic resonance imaging of 221 volunteers aged from five months to 89 years, came to the conclusion that to determine dystopia of the cerebellar tonsils, it is necessary to take

into account their age-dependent changes in position: from 1 year to 10 years - below the level of the foramen magnum by 6 mm, from 11 to 30 years - by 5 mm; from 31 years to 80 years - by 4 mm, from 81 years and beyond - by 3 mm. In 2001, R. S. Tubbs proposed to distinguish "Chiari zero malformation" - based on the presence of hydromyelia without descent of the cerebellar tonsils with a reduced volume of the posterior cranial fossa [15]. A. V. Kholin and colleagues believe that, taking into account clinical data, it is appropriate to distinguish two variants of Arnold-Chiari malformation [16]. E. G. Potemkina, based on the studies performed, supplemented the existing classifications by the degree of descent of the cerebellar tonsils into the foramen magnum and caudal: 0 degree - the tonsils are located at the level of the upper edge of the foramen magnum; 1 degree - the cerebellar tonsils are displaced to the level of the upper edge of the atlas arch; 2 degree - to the level of the upper edge of the body of the C2 vertebra; 3 degree - to the level of the upper edge of the C3 vertebra and lower in the caudal direction [17]. The most widespread at present is the classification of the American Association of Neurological Surgeons (1997), according to which three types of Arnold-Chiari malformation are distinguished: I. Posterior cerebral hernia (tonsils + - trunk) without lumbar myelomeningocele or occipitocervical encephalocele: - congenital; – acquired as a result of basal arachnoiditis, lumboperitoneal shunting, multiple lumbar punctures, II. With lumbar myelomeningocele III. With occipitocervical encephalocele [8]. However,Despite the widespread use of modern neuroimaging techniques, the etiology and pathogenesis of craniovertebral anomalies remain unclear, as does their relationship with clinical manifestations [3, 7, 8, 9]. H. Chiari was the first to put forward the theory of primary hydrocephalus in 1894. He explained the herniation of the hindbrain by intraembryonally occurring hydrocephalus, which in turn causes dystopia of the medulla oblongata and cerebellum in the caudal direction through a mechanism similar to that causing the involvement of the cerebellar tonsils in the syndrome of "intracranial hypertension". The cause of this hydrocephalus is blockage of the cerebral aqueduct. This assumption is contradicted by the fact that in most cases of Arnold-Chiari malformation the cerebral aqueduct remains normal,

and, moreover, hydrocephalus is not always detected [9]. Later, another theory, known in the literature as the "traction theory," gained recognition among some scientists. This hypothesis is based on the frequent combination of a low position of the medullary conus, a short cauda equina, and spina bifida with the Arnold–Chiari malformation. Abnormal fixation of the spinal cord in its lower sections prevents a gradual (as it grows) relative displacement in the oral direction; unable to rise upward, the spinal cord pulls the higher sections into the spinal canal cavity [10]. According to J. Cleland, the Arnold–Chiari malformation is caused by a disturbance in the formation of the brainstem during embryogenesis. A number of researchers (Barry A., Patten B., Stewart B., 1957) were supporters of the theory of disproportionate growth of the neural tube ("Disorganized neural tube growth theory"). In 1978, thanks to the studies of H. Nyland and K. Krogness, who described reduced sizes of the posterior cranial fossa in five patients with Arnold–Chiari malformation, the hypothesis about the significance of delayed growth of the skull bones compared to the development of the brain matter was confirmed [18]. Later, M. Marin-Padilla, TM Marin-Padilla conducted an experiment (1981): they administered high doses of retinol to hamsters in the early stages of pregnancy, which led to a developmental disorder of the occipital bone at the stage of embryogenesis, which subsequently contributed to a decrease in the size of the posterior cranial fossa. In the postnatal period, due to the growth of the cerebellum, the caudal part shifted downwards. Thus, the Arnold–Chiari malformation was modeled and it was proven that axial skeletal dysraphic disorders are primary in this pathology [11]. N. E. Krupina (2003), having analyzed the morphometric parameters of magnetic resonance imaging of the brain and spinal cord, craniograms and spondylograms in 364 patients with Arnold–Chiari malformation type I, revealed a frequent combination of this pathology with bone anomalies: 147 patients were diagnosed with craniosynostosis, 94 with basilar impression, 68 with platybasia, 40 with assimilation of the atlas, all patients had a reduced height of the posterior cranial fossa (Claus index),The cranial base angle is increased. After analyzing the obtained results, the researcher concluded that the bony anomalies of the vault and base of the

skull are primary in relation to Arnold–Chiari malformation type I, they lead to a decrease in the size of the posterior cranial fossa. Studying the ratio of the brainstem and cerebellum volume to the volume of the posterior cranial fossa in patients with Arnold–Chiari malformation type I, M. Nishikawa et al. (1997) discovered a disproportion in the form of an increase in this ratio, narrowing of the cerebrospinal fluid pathways, and a higher and steeper attachment of the tentorium cerebelli, which led to the conclusion about a developmental disorder of the occipital bone with the subsequent formation of a "tight" posterior cranial fossa. TH Milhorat (1999) shares this opinion after studying the morphometric parameters of the posterior cranial fossa in 364 patients. In 2001, Yu.S. Shchigolev and S.N. Larionov put forward their theory on the formation of osteoneural disproportion in Arnold–Chiari malformation type I, which is based on birth trauma. It is known that Arnold–Chiari malformation type I is often combined with hydromyelia, but the data on the frequency are contradictory: from 40 to 90% [2,7,8] (Fig. 3). Despite modern diagnostic methods, the causes of the occurrence and progression of hydromyelia in Arnold–Chiari malformation remain controversial to this day.

Conclusion. The study of craniovertebral anomalies and Arnold–Chiari malformation represents a remarkable journey from early anatomical descriptions to modern multidisciplinary analysis involving embryology, radiology, and genetics. Over centuries, researchers have progressively refined the understanding of the morphogenesis, classification, and pathophysiological mechanisms underlying these disorders. Historical data indicate that bone malformations such as basilar impression, platybasia, and atlas assimilation play a crucial role in the formation of posterior cranial fossa deformities that contribute to cerebellar tonsil descent.

Despite significant advances in diagnostic imaging, particularly MRI, the etiology and pathogenesis of Arnold–Chiari malformation remain subjects of scientific debate. Multiple theories — including those of primary hydrocephalus, traction mechanisms, and osteoneural disproportion — highlight the multifactorial nature of this condition. Modern classifications have helped systematize the complexity of

craniovertebral junction anomalies, yet no single hypothesis can fully explain all clinical and radiological manifestations.

Further interdisciplinary research combining neuroimaging, morphometric analysis, and molecular genetics is essential to clarify the embryological mechanisms and to improve diagnostic precision, prognostic assessment, and surgical planning. Understanding the interplay between structural bone development and neural tissue adaptation remains key to effective management and prevention of neurological complications associated with craniovertebral dysplasia and Arnold–Chiari malformation.

References:

1. Meadows, J., Kraut, M., Guarnieri, M., Haroun, R. I., & Carson, B. S. (2000). Asymptomatic Chiari Type I malformations identified on magnetic resonance imaging. *Journal of Neurosurgery*, *92*(6), 920–926.

2. Nishikawa, M., Sakamoto, H., Hakuba, A., Nakanishi, N., & Inoue, Y. (1997). A morphometric study of the posterior cranial fossa in Chiari malformation. *Journal of Neurosurgery*, *86*(1), 40–47.

3. Elster, A. D., & Chen, M. Y. (1992). Chiari I malformations: Clinical and radiologic reappraisal. *Radiology*, *183*(2), 347–353.

4. Mikulis, D. J., Diaz, O., Egglin, T. K., & Sanchez, R. (1992). Variance of the position of the cerebellar tonsils with age: Preliminary report. *Radiology*, *183*(3), 725–728.

5. Milhorat, T. H., et al. (1999). Chiari I malformation redefined: Clinical and radiographic findings for 364 symptomatic patients. *Neurosurgery, 44*(5), 1005–1017.

6. Pang, D., & Thompson, D. N. (2011). Embryology and bony malformations of the craniovertebral junction. *Child's Nervous System, 27*(4), 523–564.

7. Menezes, A. H. (1997). Craniovertebral junction anomalies: Diagnosis and management. *Seminars in Pediatric Neurology, 4*(3), 209–223.

8. Menezes, A. H. (2008). Craniovertebral junction database analysis: Incidence, classification, presentation and treatment algorithms. *Child's Nervous System, 24*(10), 1101–1108.

9. Smith, B. W., et al. (2013). Distribution of cerebellar tonsil position: Implications for understanding Chiari malformation. *Journal of Neurosurgery, 119*(3), 812–819.

10. Tam, S. K. P., et al. (2022). Assessment of patients with a Chiari malformation type I. *Clinical Imaging, 82*, 195–205.

11. Lawrence, B. J., et al. (2018). Cerebellar tonsil ectopia measurement in Type I Chiari malformation: Inter-operator reliability. *Journal of Clinical Imaging Science, 8*, 39.

12. Bagci, A. M., et al. (2013). Automated posterior cranial fossa volumetry by MRI. *American Journal of Neuroradiology, 34*(9), 1758–1763.

13. Botelho, R. V., et al. (2020). Posterior fossa dimensions of Chiari malformation patients: A systematic review. *World Neurosurgery, 140*, e450–e458.

14. Lloyd, R. A., et al. (2022). Statistical shape models of the posterior cranial fossa and Chiari I malformation. *Journal of Biomechanics, 141*, 111220.

15. Amer, T. A., et al. (1997). Chiari malformation type I: A new MRI classification. *Clinical Radiology, 52*(4), 270–275.

16. Brown, S. (2016). *Chiari I: Well known, not well recognized* (Radiology teaching file). University of Florida.

17. Menezes, A. H. (2012). Craniovertebral junction abnormalities with hindbrain herniation and syringomyelia. *Journal of Neurosurgery, 116*(2), 301–309.

18. Kular, S., & Nadkarni, T. (2024). Chiari Malformation Type 1. In *StatPearls*. Treasure Island (FL): StatPearls Publishing.

19. Medscape. (2024). *Chiari I Malformation (CMI) Imaging*.

20. Azarkar, G., et al. (2023). Assessing the prevalence of ectopic cerebellar tonsils in a headache cohort. *Journal of the Belgian Society of Radiology, 107*(1), 37.

21. Яриков, А. В., Леванов, В. М., & Авксентьев, Н. А. (2022). Мальформация Арнольда–Киари: этиология, патогенез, диагностика и хирургическое лечение (обзор). *Врач, 33*(4), 39–46.

22. Дзяк, Л. А., & др. (2001). Мальформация Арнольда–Киари: классификация, этиопатогенез, клиника, диагностика. *Нейрохирургия*, (3), 42–51.

23. Лобзин, С. В., & др. (2014). Краниовертебральные аномалии: принципы систематизации, теории возникновения, клинические проявления (обзор литературы). *Вестник Северо-Западного государственного медицинского университета им. И. И. Мечникова, 6*(1), 87–97.

24. Руководство (Рекомендации). (без даты). *Хирургическое лечение мальформации Киари у взрослых*. Российское общество специалистов по нейрохирургии позвоночника (пер. материалов).

25. Крупина, Н. Е., & Белодед, В. М. (2003). МРТ-особенности задней черепной ямки при семейных формах мальформации Киари. *Неврологический журнал*, (1), 24–27. (описание и ссылки в обзорах)

26. Кузбеков, А. Р., & др. (2011). Современные представления о мальформации Киари. *Вестник КазНМУ*, (спецвыпуск), 130–137. (доступ через CyberLeninka)

27. Antonenko, L. M., et al. (2015). Посттравматическое доброкачественное позиционное головокружение у пациентки с аномалией Киари I типа. *Неврологический журнал, 20*(1), 34–41. (указание в библио-каталоге)

28. *O‘zbekiston Tibbiyot Jurnali* (ред. колл.). (2017). Вып. 6(3). Ташкент: МЗ РУз (журнал в перечне ВАК; тематические обзоры по нейронаукам встречаются в номерах).

29. *Nazariy va klinik tibbiyot* (Теоретическая и клиническая медицина). (1999–н.в.). Академия наук РУз. (профильный узбекский рецензируемый журнал; пригоден для локальных обзоров по нейрохирургии/неврологии).

30. Apollo Hospitals (Uzbek). (n.d.). *Arnold–Chiari malformatsiyasi: turlari, sabablari, belgilari va davolash*. Обзор для пациентов на узбекском языке (для локального цитирования терминологии и популяризаторских описаний).

31. Abduvoyitov, B. (2025). FARMAKOREZISTENT EPILEPSIYA BILAN OG'RIGAN BEMORLARNING JARROHLIK DAVOLASH NATIJALARI VA QONIQARSIZ JARROHLIK NATIJALARINI BASHORAT QILUVCHI OMILLAR. *Modern Science and Research*, *4*(2), 91-92.

32. Abduvoyitov, B. (2025). POLIMIKROGIRIYALI FARMAKOREZISTENT EPILEPSIYANI DAVOLASH NATIJALARI. *Modern Science and Research*, *4*(2), 117-121.

33. Abduvoyitov, B. (2025). FARMAKOREZISTENT EPILEPSIYADA STEROIDLARNING AHAMIYATI. *Medical Research Journal*, *1*(1), 244-250.

34. Abduvoyitov, B. (2025). MRT ASOSIDA FARMAKO REZITENT EPILEPSIYANI TASHXISLASH VA NATIJALARNI PROGNOZ QILISH: ZAMONAVIY YONDASHUVLAR. *Medical Research Journal*, *1*(1), 215-219.

35. Мамадалиев, А. М., Алиев, М. А., Абдувойитов, Б. Б. У., Хайритдинов, Б. Б., Фарухова, М. Ф., Гаппарова, О. И., ... & Бурхонов, А. Ш. (2022). Клинический случай риносинусогенного абсцесса головного мозга и обзор литературы. *Uzbek journal of case reports*, *2*(2), 7-11.

36. Bahodirovich A. B. et al. Approaches to intestinal decompression during different appendicular peritonitis in children //Достижения науки и образования. – 2018. – №. 18 (40). – С. 92-95.

37. Djalolov D. A. et al. Features of microflora in the etiological structure of diffuse appendicular peritonitis //Вопросы науки и образования. – 2018. – Т. 8. – №. 2. – С. 116.

38. Abduvoyitov Bobur Bahodirovich, Djalolov Davlatshokh Abduvokhidovich, Khasanov Aziz Batirovich, Abbasov Khojimuhammad Khabibullayevich The effect of ozone on the course and development of complications of peritonitis in children // Вопросы науки и образования. 2018. №29 (41).

39. Abduvoyitov B. B. et al. The effect of ozone on the course and development of complications of peritonitis in children //Вопросы науки и образования. – 2018. – Т. 29. – С. 110-113.

40. Bahodirovich A. B. et al. Immunological feature of the body in children with diffused appendicular peritonitis //Вопросы науки и образования. – 2019. – №. 2 (45). – С. 115-122.