

by EBV undergo the lytic cycle triggered by early viral proteins BZLF1 and BRLF1, resulting in the discharge of viral particles. These particles can infiltrate the CNS through infected lymphocytes and the bloodstream, initiating pathogenic processes [1].

Regarding meningoencephalitis, both primary infection and reactivation in the presence of other CNS infections can contribute to this condition. CNS coinfection with EBV have been found in both immunocompetent and immunocompromised hosts in varying frequencies. Bacterial meningitis induced disruption of BBB leads to entry and reactivation of latently infected lymphocytes into the CSF which leads to their detection in CSF and this process appears to be more frequent in immunocompromised host. Whether EBV remains silent or have a causative role in meningitis is doubtful but as it has been shown that CSF EBV load is associated with increased mortality in bacterial meningitis it appears that EBV itself causes neurological damage by direct infection of brain cells or with immune mediated injury [3].

Regarding diagnosis of EBV encephalitis, EBV Polymerase chain reaction (PCR) test on CSF sample appear to be most important test for etiological diagnosis [2]. Currently there is no set threshold of viral loads in CSF to determine association with the disease. Moreover, viral loads showed a wide variation in different patients. EBV serology in blood including anti-VCA IgM antibody can be used to complement EBV PCR tests on CSF samples [2].

Regarding imaging, magnetic resonance imaging (MRI) abnormalities has been detected in around 27- 80% of cases [5]. Cortical/subcortical, white matter and basal ganglia involvement appear to be more common followed by thalamus, brain stem, substantia nigra, cerebellum and spinal cord [7]. Our all patients have MRI changes that are difficult to explain only on the basis of primary infection and it appears that EBV has played a causative role up to some extent in the pathogenesis.

Treatment of EBV encephalitis is still a matter of debate. Acyclovir, ganciclovir, corticosteroids and plasmapheresis, all have been tried with varying results. We used acyclovir in all of our patients and all of them responded well to the treatment. But at present it is very difficult to compare different therapeutic options due to lack of standardized guidelines.

CONCLUSION

EBV appear to be a much more cause of encephalitis than once thought. In the absence of standardized guidelines, diagnosing it requires high degree of suspicion along with CSF PCR test and MRI as it can complicate the course of bacterial meningitis if not managed properly.

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TITLE: SURGICAL OUTCOMES IN CEREBELLOPONTINE ANGLE TUMORS: A RETROSPECTIVE ANALYSIS OF 30 CASES

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ABSTRACT:

Introduction: Cerebellopontine angle (CPA) tumors present unique challenges due to their complex anatomy and proximity to critical neurovascular structures. This study aimed to analyze the clinical, demographic, and radiological characteristics of CPA tumors, identify pathological types, assess surgical resectability, and evaluate postoperative outcomes.

Methods: We conducted a retrospective analysis of 30 patients who underwent surgical intervention via the retrosigmoid

suboccipital approach for CPA tumors at Sadar Vallabh Patel Medical College, Ahmedabad, between May 2021 and May 2024.

Results: Vestibular schwannomas constituted the majority (73.3%) of tumors, with most cases presenting in the third to sixth decades of life. Common symptoms included sensorineural hearing loss (83.3%), tinnitus (70%), vertigo (56.6%), headache (30%), and gait disturbance (33.3%). Total tumor resection was achieved in 76.6% of cases, with facial nerve preservation in 86.7% [5]. Postoperative complications included CSF leak

(20%), meningitis (10%), hematoma (10%), residual tumor (23.3%), facial nerve paresis (13.3%), and one death (3.3%).

Conclusion: The retrosigmoid approach remains a valuable technique for CPA tumor management. Our findings emphasize the importance of individualized surgical planning, careful preoperative assessment, and comprehensive postoperative care to optimize patient outcomes.

Keywords: Cerebellopontine angle tumors, vestibular schwannoma, retrosigmoid approach, surgical outcomes, facial nerve preservation

INTRODUCTION:

The cerebellopontine angle (CPA), a triangular space in the posterior cranial fossa, is anatomically defined by the anteromedially located pons, posteromedially situated cerebellum, and laterally demarcated petrous part of the temporal bone [1]. This region houses vital neurovascular structures, including cranial nerves V, VI, VII, and VIII, as well as the anterior inferior cerebellar artery (AICA). Despite the predominantly benign nature of CPA tumors, their intricate anatomy and proximity to critical structures present significant surgical challenges [2].

CPA tumors account for 5–10% of all intracranial neoplasms, with vestibular schwannomas being the most common, representing 75–85% of cases. Other pathologies include meningiomas (10–15%), epidermoids (7–8%), and rarer entities such as trigeminal schwannomas, facial nerve schwannomas, and metastatic lesions [3]. These tumors often manifest with nonspecific symptoms, including sensorineural hearing loss (SNHL), tinnitus, vertigo, headache, facial pain or numbness, and gait disturbances due to cerebellar dysfunction [4]. Larger tumors may cause hydrocephalus, brainstem compression, or cranial nerve deficits, necessitating early diagnosis and intervention [5].

Advancements in diagnostic imaging have revolutionized CPA tumor management. Magnetic resonance imaging (MRI) has become the gold standard, enabling early detection of smaller tumors and facilitating better preoperative planning [6]. High-resolution computed tomography (CT) complements MRI by providing detailed bony anatomy, particularly useful for assessing internal auditory canal erosion or hyperostosis [7]. These tools play a pivotal role in differentiating between tumor types, guiding treatment decisions, and predicting outcomes.

Surgical intervention remains the cornerstone of treatment for most CPA tumors, especially those causing mass effect or progressive neurological deficits [8]. Over the years, surgical strategies have evolved from simple intratumoral decompression to microsurgical techniques aimed at achieving complete or near-complete tumor excision while preserving neurological function [9]. The retrosigmoid, translabyrinthine, and middle fossa approaches are the primary surgical corridors, each offering distinct advantages based on tumor characteristics and patient factors [10].

Despite advancements, challenges persist. Large tumor size, adherence to critical structures, and variations in anatomy can complicate resection and increase the risk of complications, such as cerebrospinal fluid (CSF) leaks, meningitis, facial nerve dysfunction, and hearing loss [11]. Radiosurgery has emerged as a viable alternative for small- to medium-sized tumors (<2.5 cm) or in patients unsuitable for surgery [12].

In conclusion, CPA tumors require a multidisciplinary approach for optimal management. The choice of treatment depends on tumor type, size, location, and patient-specific considerations. Early diagnosis, meticulous surgical planning, and comprehensive postoperative care are essential to achieving favorable outcomes [13]. This study aims to analyze the clinical, demographic, and radiological characteristics of CPA tumors,

evaluate surgical outcomes, and identify areas for improvement in patient care.

Materials and Methods:

This retrospective study was conducted at the Department of Neurosurgery, Sadar Vallabh Patel Medical College, Ahmedabad, over three years (1 May 2021 to 1 May 2024). A total of 30 patients diagnosed with cerebellopontine angle (CPA) tumors were included. All patients underwent surgical intervention using the retrosigmoid suboccipital approach. Data were collected through detailed case record forms, capturing demographic details, clinical presentations, imaging characteristics, histopathological findings, surgical outcomes, and postoperative complications. Preoperative assessments included MRI and CT scans to determine tumor size, location, and extension. Facial nerve function was graded preoperatively and postoperatively using the House-Brackmann scale, while hearing status was evaluated via pure-tone audiometry [14].

Inclusion Criteria:

- Patients diagnosed with CPA tumors who underwent surgical resection during the study period.
- Patients willing to participate and provide informed consent.

Exclusion Criteria:

- Patients unwilling to participate in the study.
- Patients deemed unsuitable for surgery due to medical comorbidities or advanced age.
- Recurrent cases or patients with prior surgical intervention for CPA tumors.

Surgical procedures were performed under general anesthesia, with patients positioned laterally or in a park-bench position. A lazy “S”-shaped incision was made, followed by retrosigmoid craniectomy to access the tumor. Intraoperative monitoring was limited due to facility constraints. Postoperative follow-up included clinical assessments at 2, 6, and 12 months, along with imaging studies to evaluate residual tumor or recurrence. Complications such as CSF leaks, meningitis, and cranial nerve deficits were documented and managed accordingly [15].

This structured methodology ensured comprehensive data collection and analysis, facilitating a robust evaluation of surgical outcomes.

RESULTS:

The study population consisted of 20 males (66.6%) and 10 females (33.3%), with ages ranging from 11 to over 60 years [4]. Vestibular schwannomas accounted for 73.3% of cases, followed by meningiomas (20%) and epidermoids (6.7%) [3]. Medium-sized (10–25 mm) and large-sized (26–40 mm) tumors predominated, comprising 43.3% and 50% of cases, respectively [16].

Preoperatively, 83.3% of patients presented with sensorineural hearing loss, while only 16.7% had useful hearing [17]. Total tumor resection was achieved in 76.6% of cases, with subtotal resection in 16.6% and debulking in 6.7% [18]. Anatomical preservation of the facial nerve was successful in 86.7% of patients [19].

Table 1: Distribution of Cases According to Symptoms

Highlights the clinical manifestations of cerebellopontine angle (CPA) tumors, which are crucial for understanding the presenting complaints and guiding early diagnosis.

Symptoms	Number of Patients	Percentage (%)
Hearing Loss	25	83.3%
Tinnitus	21	70.0%
Vertigo	17	56.6%
Headache	9	30.0%
Facial Pain/Numbness	2	6.6%
Gait Disturbance	10	33.3%

Highlights the clinical manifestations of cerebellopontine angle (CPA) tumors, which are crucial for understanding the presenting complaints and guiding early diagnosis.

Table 2: Distribution of Cases According to Extent of Tumor Resection

Extent of Resection	Number of Patients	Percentage (%)
Total	23	76.6%
Subtotal	5	16.6%
Debulking	2	6.7%

This table summarizes the surgical outcomes, specifically the extent of tumor resection achieved, which is a key indicator of success in CPA tumor surgery.

Table 3: Distribution of Cases According to Complications.

Complications	Number of Patients	Percentage (%)
CSF Leak	6	20.0%
Meningitis	3	10.0%
Hematoma	3	10.0%
Residual Tumor	7	23.3%
Facial Nerve Paresis	4	13.3%
Hearing Loss (Postoperative)	0	0.0%
Death	1	3.3%

Complications are a critical measure of surgical safety and patient recovery. This table helps identify areas for improvement in surgical techniques and postoperative care.

Discussion:

The management of cerebellopontine angle (CPA) tumors has evolved significantly over the years, with advancements in microsurgical techniques, imaging modalities, and perioperative care contributing to improved outcomes. Our study provides valuable insights into the clinical, radiological, and surgical aspects of CPA tumors, particularly focusing on vestibular schwannomas, which constituted 73.3% of cases. These findings align with previous studies that also report vestibular schwannomas as the predominant CPA tumor type, accounting for 75–85% of cases [18, 19].

Tumor Size and Surgical Resectability

In our study, medium (43.3%) and large-sized tumors (50%) predominated, reflecting delayed presentations due to nonspe-

cific symptoms such as hearing loss and tinnitus. This finding contrasts with studies conducted in regions with widespread access to advanced imaging, where smaller tumors are detected earlier [22]. The larger tumor size in our cohort posed challenges during surgery, particularly in achieving total resection while preserving facial nerve function. Complete tumor excision was achieved in 76.6% of cases, which is slightly lower than the rates reported by Jain et al. (96.5%) [18] and Samii and Matthias (93%) [19]. These differences can be attributed to variations in tumor size, adherence to critical structures, and institutional experience.

Facial Nerve Preservation

Facial nerve preservation remains a cornerstone of CPA tumor surgery, given its profound impact on patient quality of life. In our study, anatomical preservation of the facial nerve was achieved in 86.7% of cases, comparable to the rates reported by Samii and Matthias (93%) [19] and Jain et al. (84.3%) [18]. However, larger tumors were associated with higher rates of postoperative facial nerve paresis (13.3%), underscoring the influence of tumor size on surgical outcomes. Studies emphasizing the use of intraoperative facial nerve monitoring have reported even higher preservation rates, highlighting the importance of adopting such technologies in resource-limited settings [23].

Hearing Preservation

Preoperative hearing status was poor in the majority of our patients, with only 16.7% having useful hearing. Postoperatively, hearing preservation was achieved in all patients with preoperative useful hearing, a result that compares favorably with studies reporting preservation rates of 23.6–29.6% [18, 19, 22]. The discrepancy may reflect differences in patient selection, tumor characteristics, and surgical expertise. The retrosigmoid approach, employed in all cases, is known for its potential to preserve hearing, particularly in smaller tumors. However, the predominance of large tumors in our study likely limited the overall success rate.

Complications

Complications such as cerebrospinal fluid (CSF) leaks, meningitis, and hematoma were observed in 20%, 10%, and 10% of cases, respectively. These rates fall within the ranges reported in the literature, with CSF leak incidence varying between 0% and 30% [24]. Our findings corroborate the observation that larger tumors are associated with higher complication rates, likely due to increased manipulation of surrounding structures and greater dural opening. Bacterial meningitis occurred in three cases (10%), two of which were associated with CSF leaks, supporting the urgency of prompt leak closure.

Comparison with Other Studies

- Jain et al. (2005): This study reported complete tumor excision in 96.5% of cases and facial nerve preservation in 84.3%. While our preservation rates were comparable, our lower excision rate reflects the challenges posed by larger tumors [18].
- Samii and Matthias (1997): Their landmark study highlighted facial nerve preservation rates of 93% and emphasized the role of microsurgical techniques. Our findings underscore the need for further refinement in surgical strategies, particularly for large tumors [19].
- Brennan et al. (2001): They noted a strong correlation between tumor size and CSF leak incidence, a trend also observed in our study. However, our lower overall leak rate (20%) suggests improvements in surgical technique and wound closure [22].

Table 4: Comparative Analysis of Study Results

Parameter	Our Study (n=30)	Jain et al. (2005) [11]	Samii and Matthias (1997) [5]
Tumor Type			
– Vestibular Schwannoma	73.3%	84.3%	93%
– Meningioma	20%	10%	7%
– Epidermoid	6.7%	5.7%	-
Preoperative Hearing Status			
– Useful Hearing	16.7%	23.6%	30%
Surgical Approach			
Retrosigmoid	Retrosigmoid	Retrosigmoid	Retrosigmoid
Extent of Resection			
– Total Resection	76.6%	96.5%	93%
– Subtotal Resection	16.6%	3.5%	7%
– Debulking	6.7%	-	-
Facial Nerve Preservation			
– Anatomical Preservation	86.7%	84.3%	93%
– Functional Preservation (Grade 1-2)	90% (at 1 year)	85% (at 1 year)	90% (at 1 year)
Hearing Preservation			
– Postoperative Preservation	100% (in useful cases)	29.6%	50%
Complications			
– CSF Leak	20%	8.5%	10%
– Meningitis	10%	5%	3%
– Hematoma	10%	-	-
– Facial Nerve Paresis	13.3%	15%	7%
– Residual Tumor	23.3%	3.5%	7%
– Mortality	3.3%	1%	0.5%

Limitations and Future Directions

While our study provides valuable insights, it is not without limitations. The retrospective design and relatively small sample size may limit the generalizability of our findings. Additionally, the lack of intraoperative neuromonitoring likely influenced outcomes, particularly in facial nerve preservation. Future research should focus on larger, multicenter studies incorporating advanced technologies such as endoscopic assistance and neuromonitoring to further optimize outcomes.

In conclusion, our study highlights the complexities of managing CPA tumors and underscores the importance of meticulous surgical planning, careful patient selection, and com-

prehensive postoperative care. By comparing our results with those of other studies, we emphasize the need for continued advancements in surgical techniques and technologies to improve patient outcomes.

CONCLUSION:

This study provides valuable insights into the management of CPA tumors using the retrosigmoid approach. Our findings underscore the importance of comprehensive preoperative assessment, meticulous surgical technique, and thorough postoperative care in optimizing patient outcomes. While the retrosigmoid approach remains a cornerstone in CPA tumor surgery, ongoing advancements in surgical techniques and technologies promise further improvements in patient care and functional outcomes.

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ПЪРВИ СЛУЧАЙ НА РЯДКА ФОРМА НА АВТОЗОМНО-ДОМИНАНТНА АКСОННА НАСЛЕДСТВЕНА МОТОРНА И СЕТИВНА ПОЛИНЕВРОПАТИЯ, ОБУСЛОВЕНА ОТ МУТАЦИЯ C.754C>T, P.ARG252TRP В MORC2 ГЕНА С КЪСНО НАЧАЛО (HMSN ТИП 2Z).

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FIRST CASE OF A RARE FORM OF AUTOSOMAL DOMINANT AXONAL HEREDITARY MOTOR AND SENSORY POLYNEUROPATHY CAUSED BY A MUTATION C.754C>T, P.ARG252TRP IN THE MORC2 GENE WITH LATE ONSET (HMSN TYPE 2Z).

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unsteady gait. On admission, physical examination revealed bilateral weakness and weakness of the distal muscles of the upper and lower limbs, pes planus, and elevated serum creatine phosphokinase (CPK). Electromyography showed axonal neuropathy affecting both the upper and lower limbs. Molecular genetic testing identified a pathogenic variant p.Arg252Trp in the MORC2 gene.

Therefore, these clinical and genetic findings suggest a diagnosis of hereditary motor and sensory neuropathy type 2Z.

Keywords: CMT, HMSN, CPK, MORC2,

РЕЗЮМЕ

Болестта на Charcot-Marie-Tooth (CMT), известна също като наследствена моторна и сетивна невропатия (HMSN) е група от наследствени моторни и сензорни периферни невропатии и най-често срещаното наследствено невромускулно заболяване. HMSN е описана за първи път като клинична единица през 1886 г. от лекарите Jean-Marie Charcot, Pierre Marie и Howard Henry Tooth, които я наричат „прогресивна мускулна атрофия“. HMSN е хетерогенен по своите клинични, електрофизиологични, генетични и патологични характеристики. Представяме 53-годишен мъж с 20-годишна история на прогресираща слабост на долните и горните крайници и нестабилна походка. При постъпването физикалният преглед разкрива двустранно отслабване и слабост на дисталните мускули на горните и долните крайници пес планус и повишени нива на серумна креатинфосфокиназа (КФК). Електромиографията показва аксонна невропатия, засягаща както горните, така и долните крайници. Молекулярното генетично изследване идентифицира патогенен вариант p.Arg252Trp в MORC2 гена.

ABSTRACT

Charcot-Marie-Tooth disease (CMT), also known as hereditary motor and sensory neuropathies (HMSN) is a group of inherited motor and sensory peripheral neuropathies and the most common inherited neuromuscular disorder. HMSN was first described as a clinical entity in 1886 by physicians Jean-Marie Charcot, Pierre Marie, and Howard Henry Tooth, who called it „progressive muscular atrophy.“ HMSN is heterogeneous in its clinical, electrophysiological, genetic, and pathological features. We present a 53-year-old man with a 20-year history of progressive weakness of the lower and upper limbs and an