CASE REPORT

An unusual case of a mixed germ cell tumour presenting with complete blindness

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Keywords: Non-germinomatous germ cell tumours, intracranial germ cell tumours, mixed germ cell tumours, Neuro-oncology, Case Report

Introduction

Primary intracranial germ cell tumours (IGCTs) comprise of 0.43% to 0.54% of all CNS tumours with an incidence of 0.10 per 100,000 person years and broadly classified as pure germinomatous (PG) (70-80%) and non-germinomatous germ cell tumours (NGGCTs)(20-30%).[1,2] This study reports the first known case in Sri Lankan literature of a paediatric patient with a mixed germ cell tumour(MGCT), a type of NGGCT, with an unusual presentation of complete vision loss.

Case study

A 15-year-old female presented with a 2-week history of sudden onset of bilateral loss of vision associated with a diffuse bilateral headache in the background of low mood, irritability, and anhedonia and anergia for 3 months managed as depression with oral antidepressants. On admission, she was drowsy with a GCS of 13/15 (E1, V5, M6) and a neurological examination demonstrated complete visual blindness. A NCCT brain that followed demonstrated a sella-suprasellar lesion causing hydrocephalus. Blood investigations revealed normal serum electrolytes, FSH, LH, cortisol levels but an elevated prolactin level of 1172 mIU/L.

MRI scan of the brain showed a large supra sellar lesion suggestive of an optic chiasmatic glioma (volume of 22.457 cm^3), displaying heterogenous high signal intensity on DW and T2 weighted images and high signal intensity on FLAIR images with strong contrast enhancement suggestive of a glial tumour. The tumour was seen extending anteriorly to the optic chiasm and posteriorly along the 3rd ventricle and towards the mid brain. (Figure 1)

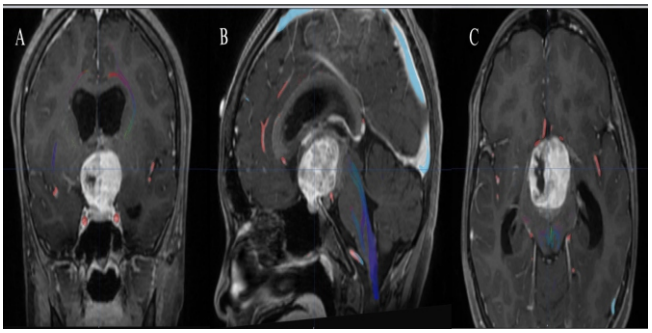

Figure 1. (a) Coronal; (b) Sagittal; (c) Axial Neuro-navigation MRI demonstrating a large mass lesion in the suprasellar region with contrast enhancement extending posteriorly to the mid-brain and anteriorly to the optic chiasm. (Volume 22.437 cm^3) (Overlays demonstrate arteries (red), veins (blue) and tracts/fibres (colour spectra))

She was initially stabilized using Levatiracetam, mannitol and methyl prednisolone and underwent neuro-navigation guided total excision using a pterional approach and placement of a right sided medium pressure ventriculoperitoneal shunt, where total excision was achieved while preserving the optic nerves and the carotid arteries.

Histology revealed tumour sections with mixed morphological patterns of teratoma, yolk sac and germinomas. Immunohistochemistry showed positivity for CD 117 (40% in germinomatous areas), alpha-feto protein (AFP) (40% in yolk sac elements), CD 30 and EMA (occasional staining). Accordingly, an immunomorphological diagnosis of a mixed germ cell tumour (germinoma, yolk sac and immature teratomatous components) was made.

At 1 month of follow-up the patient had acquired complete back and white vision in both eyes and at three months recovered complete colour vision in the left eye and partial vision in the right eye. However, she did not undergo chemoradiotherapy postoperatively due to refusal from the guardian in fear of side effects.

Discussion

MGCTs, a rare entity accounting for 1.69-10.83% of ICGSTs and classified under NGGCTs are defined according to the

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https://orcid.org/0000-0001-7879-9439
Received: 20-04-2024 Accepted: 01-12-2024
DOI: https://doi.org/10.4038/sljs.v42i3.9136

2021 WHO Classification of CNS Tumours as malignant GCTs harbouring at least two GCT subtypes in any combination. [1,2] Accounting for a male preponderance like other GCTs, MCGTs are broadly classified into three types; (1) mixed germinoma and teratoma, (2) mixed tumours comprising mainly of predominant germinoma or teratoma and (3) tumours consisting of purely malignant components, out of which the current case belonged to type 1. [2] This type is considered under the intermediate prognostic group based on the therapeutic classification of IGCTs with overall 3- and 5-year OS of 94.1% and 84.7% respectively. [2,3]

While the lesion in the present case was in the suprasellar area which is also the second commonest site of IGCTs (22%) surpassed only by the pineal region accounting for 53%, serum markers of β hCG and AFP were not carried out due to the presumptive radiological diagnosis of a glioma.[2] This was further justified by the atypical presentation of the current patient, unlike the usual presenting complaints of headache, vomiting or features of diabetes insipidus which are commonly associated with IGCTs. [3-5] This is in keeping with current literature, where imaging and clinical characteristics of MGCTs can vary widely pertaining to the mixed components of the tumour.[4]

Management of MGCTs are still without consensus owing to its rarity and different studies have depicted the utilization of different treatment strategies. However, it is in agreement that aggressive treatment with biopsy or surgical resection in combination with adjuvant chemoradiotherapy can be utilized in various combinations in its management since all IGCTs except mature teratomas are considered malignant and neither surgery alone nor chemoradiotherapy alone is sufficient to achieve complete cure.[4] Nevertheless, it is important to understand that the best treatment regimen is still debatable with some large scale studies such as those based on the SEER database demonstrate improved survival in addition of chemotherapy to resection (over resection alone) which is in contrast to the 2013 CNS GCT Symposium recommendations of chemoradiotherapy followed by resection for residual disease, hence necessitating clinical trials to further elucidate on it.[1] On the other hand certain other studies advice 25 Gy ventricular/whole brain and 25 Gy tumour boost for localized MGCTs following biopsy or resection, with chemotherapy pre- and post-radiotherapy only if disseminated or at recurrence.[4] Hence, this lack of consensus illustrates the dire requirement of clinical trials to standardize treatment protocols.

Even though extensive resection is mainly advocated towards management of mature teratomas, surgery in NGGCTs is commonly limited to a biopsy for acquiring a histological diagnosis and in limited cases to achieve partial resection.[3] However, we believe that open surgery with an attempt to obtain maximal debulking in the hands of an experienced surgeon far outweighs the utilization of stereotactic biopsies in the suprasellar and pineal regions with its anatomically critical neurovascular neighbours particularly in instances such as the current case where there are imminent pressure effects on vital structures such as the optic chiasm causing vision abnormalities.

Conclusions

In conclusion, this is an interesting case of a paediatric MGCT presenting with complete visual loss and suggests the possible role of surgery in the management of NGGCTs. Moreover, it is an excellent example of the high degree of suspicion that ought to be present regarding intracranial space occupying lesions in the paediatric population and the importance of accurate histological diagnosis in planning postoperative oncological management.

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Learning Points:

- Clinicians should evaluate intracranial SOLs of paediatric patients with a higher degree of suspicion towards germ cell tumours.
- Histopathological diagnosis in the form of a surgical biopsy is essential in the management of intracranial germ cell tumours.
- Carefully planned large-scale studies are needed to evaluate the potential of surgery in surpassing a mere diagnostic role in the management of malignant germ cell tumours.