Radiological latency in pineal germinoma; a case report and literature review

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Male / 23 / good past health

Presented to ophthalmology with 3 months history of diplopia (since late 2017)

Assessment at the time revealed dorsal midbrain signs;

- Pupillary light-near dissociation bilaterally
- Convergence retraction nystagmus on vertical saccades
- Upgaze palsy

1/2018 MRI brain with contrast:
  No lesion

Worsening diplopia on follow up in 2019

6/2020 MRI brain with contrast:
  Pineal region tumour
On admission to Neurosurgery, also noted:

Polydipsia + polyuria → consistent with DI
Also noted low fT4

Serum and CSF tumour markers (AFP, alpha fetoprotein, human chorionic gonadotropin):
Normal

Varioguide frameless stereotactic biopsy of pineal region tumour performed

Pathology:
Pineal germinoma
Focal signs in the absence of radiological evidence

Latency of radiologically discrete lesion

So what?

This phenomenon also seen in some patients with suprasellar germinoma, presenting with symptoms of diabetes insipidus prior to radiological evidence of discrete lesions, as reported in literature...

Mootha et al [1] reported in a study of nine children, clinical latency between appearance of symptoms to a biopsy-proven diagnosis of germinoma based on initial MRI findings as seen in the table above.

In our series, 44.4% of patients with suprasellar GCTs received an initial diagnosis of idiopathic central DI because the tumour was not detectable in the initial MRI. In these patients, the mean time elapsed from the onset of symptoms with normal MRI findings to the evidence of suprasellar tumour in MRI was 21 months, which was consistent with

In another retrospective review of intracranial germ cell tumours by Carpio et al [2], a similar phenomenon was observed.
What about pineal germinoma?

A retrospective cohort study by Phi et al [3] of 181 patients with intracranial germ cell tumour, 17 patients had a delayed diagnosis of more than 90 days since initial MRI imaging.

2 of these patients who presented with precocious puberty, went on to develop pineal region germinoma. Both initially had normal MRI scans.

Another case report [4] of a 21 year old man who developed enhancing suprasellar and pretectal masses on subsequent MRI brain scans, presumed to be germinomas. Much like our patient, presented with a normal initial MRI, but florid ophthalmological signs including pupillary light-near dissociation, convergence-retraction nystagmus, and up-gaze palsy.

The phenomenon of “radiological latency” is well documented in patients with suprasellar germinoma, but the literature documenting this same phenomenon in patients with pineal region germinomas is sparse. Indeed, the evidence exists and well demonstrated in select studies / case reports, and our case characterizes this phenomenon clearly. In fact, through a review of the literature, this phenomenon can be seen in intracranial germ cell tumours of other regions as well, including the basal ganglia.

Clinical implications?

High index of suspicion in the absence of initial radiological evidence should the patient present with symptoms of diabetes insipidus, or ophthalmological signs such as those characterizing Parinaud syndrome

Adopt a follow up MRI protocol for suspected cases eg. follow up scan every 6 months in order to minimize time to diagnosis

References