CENTRAL NERVOUS SYSTEM GERMINOMA PRESENTING AS PROGRESSIVE CEREBRAL HEMIATROPHY AND PITUITARY ENLARGEMENT

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BACKGROUND

Central nervous system (CNS) germinomas account for 0.5-2.1% of intracranial tumors with a peak incidence in the second decade of life. The most common location is in midline structures, specially the suprasellar and pineal regions. Simultaneous involvement of midline and off-midline structures has only rarely been reported. We aim to describe an atypical presentation of CNS germinoma and discuss its clinical-pathological findings.

CASE REPORT

A 20-year-old, white, right-handed woman presented with a 4-year history of progressive left-sided weakness and behavior changes, evolving with focal motor seizures involving the left arm. The patient also had a history of primary amenorrhea. Neurological examination revealed left hemiparesis, left hemianopia, constructional and dressing apraxia, executive disfunction and right Horner’s. Brain MRI showed right cerebral hemiatrophy, involving basal ganglia and thalamus; a pituitary study demonstrated enlargement of the gland and heterogeneous contrast enhancement (Figure 1A-E). FDG PET-CT (Figure 1F-H) disclosed increased glycolytic metabolism in the pituitary region and hypometabolism in the right cerebral hemisphere. Blood tests, serologies and CSF analysis were unremarkable, but a hormone panel showed panhypopituitarism. Eventually a pituitary biopsy was performed. The specimen revealed large neoplastic cells arranged in sheets with infiltration of small lymphocytes (Figure 2A-F). Immunohistochemical analysis was compatible with the diagnosis of germinoma, by the WHO classification of tumours of the CNS.

DISCUSSION & CONCLUSIONS

In children and young adults, progressive cerebral hemiatrophy can occur with varying inflammatory and neoplastic diseases, including hemiconvulsion-hemiplegia-epilepsy syndrome, Rasmussen’s encephalitis, linear scleroderma, Parry-Romberg syndrome and intracranial germ cell tumors. Moreover, acquired hypopituitarism with suprasellar enhancing masses can be caused by pituitary tumors such as adenomas or germ cell tumors, inflammatory (hypophysitis, sarcoidosis) or infiltrative diseases (Langerhans cell histiocytosis). Although both cerebral hemiatrophy and pituitary masses have broad differential diagnoses, the concurrence of both disorders was key to the diagnosis in this case. Germinomas with simultaneous involvement of midline and off-midline structures have only rarely been reported.

References: