Lobar haemorrhagic mass in a young girl with neurofibromatosis type 1

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DESCRIPTION

A 9-year-old girl with neurofibromatosis type 1 (NF-1) presented with sudden onset neurological deterioration following a 2-day history of headache. She was comatose on arrival with right third nerve palsy and contralateral hemiparesis. A head CT revealed a large right frontal acute haemorrhagic lesion with midline shift and uncal herniation (figure 1). The patient underwent emergent decompressive craniotomy and resection of the mass lesion where, the pathology demonstrated a highly cellular glial tumour with extensive nuclear pleomorphism and increased mitotic index consistent with a diagnosis of high-grade glioma (figure 2).

The differential diagnosis of an acute non-traumatic cortical haemorrhage in a child with NF1 includes aneurysm, haemorrhagic transformation of an ischaemic stroke and tumour. While majority of parenchymal tumours in NF-1 are low-grade astrocytomas, the rapid progression of symptoms in association with cerebral haemorrhage supported the diagnosis of a high-grade neoplasm.

NF-1 is a genetic tumour syndrome in which the neurofibromin, one of the main Ras-GAP proteins, is silenced on chr 17q11.2. Ras-GAP proteins activate Ras-GTP-hydrolase, inactivating the Ras signalling pathway that promotes cell proliferation. While low-grade astrocytomas are most commonly associated with NF-1, they are generally not haemorrhagic. Malignant gliomas have been rarely reported in children with NF-1.1 2 Experiments in animal models suggest that early inactivation of the p53 tumour suppressor gene may act in synergy with downstream Ras pathways to promote high grade transformation.3 Our case demonstrates the rare association of high-grade tumours with NF-1 and expands the differential diagnosis of haemorrhagic lesions in the patient population.

Learning points

▸ Brain tumours in children may present with intracranial haemorrhage.
▸ High-grade glioma is a rare tumour in association with neurofibromatosis type 1 and should be included in the differential diagnosis

Competing interests None.

Patient consent Obtained.

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Figure 1 Preoperative head CT demonstrating a large right frontal lobar heterogeneous haemorrhage with acute and subacute components without oedema, an 8 mm midline shift and uncal herniation (right).

Figure 2 H&E-stained biopsy specimen demonstrates a glial tumour with extensive nuclear pleomorphism and brisk mitotic activity in a microcystic background consistent with diagnosis of a high-grade glioma.
REFERENCES

