Craniopharyngiomas are among the most common paediatric tumours and are thought to arise from embryonic remnants of Rathke’s pouch. The molecular mechanisms involved in their formation remain elusive and little is known about chromosomal imbalances that could suggest the locations of tumour suppressor or proto-oncogenes involved in the pathogenesis. The paucity of published data on the molecular basis of such tumours prompted this investigation of 20 adamantinomatous and nine papillary craniopharyngiomas for genetic abnormalities by comparative genomic hybridisation (CGH). CGH revealed no DNA copy number changes in any of the 29 primary craniopharyngiomas, regardless of their histological subtype. These data suggest that chromosomal imbalances are a rare event in both adamantinomatous and papillary craniopharyngiomas.

RESULTS

CGH revealed no DNA copy number changes in any of the 29 primary craniopharyngiomas, regardless of their histological subtype. Successful completion and the quality of CGH investigation in each case was established by checking the narrowness of the 95% confidence interval as well as the loss of the Y chromosome in tumour material from female patients hybridised on metaphase spreads of a male donor (internal positive control).
DISCUSSION
Craniopharyngiomas are benign tumours that show a bimodal age distribution and arise in two distinct clinicopathological variants: the adamantinomatous and the papillary subtypes. The molecular mechanisms involved in craniopharyngiomas remain elusive. While a genetic susceptibility is not known, there are reports describing the occurrence of craniopharyngiomas in consanguineous siblings as well as in a mother and daughter. To date, cytogenetic (that is, karyotypic) data on only 11 craniopharyngiomas have been published, and have shown multiple chromosomal abnormalities in two cases, both of which involved chromosomes 2 and 12, while the other nine cases presented with normal karyotypes; four additional craniopharyngiomas showed no mutations of the TP53 tumour suppressor gene. In view of the association of naevoid basal cell carcinoma or Gorlin syndrome with the occurrence of craniopharyngiomas, a recent study was carried out on 22 adamantinomatous craniopharyngiomas. This found no mutations in the Gorlin syndrome gene PTCH, localised on chromosome 9q22.3, while the putative proto-oncogenes encoding the α subunits of the stimulatory (Gsα) or the inhibitory (Gi2α) GTP binding proteins on respective chromosome subunits 20q13.2 and 3p21 were also found not to be mutated. Interestingly, a subset of these adamantinomatous craniopharyngiomas turned out to be monoclonal in origin.

While it has to be borne in mind that CGH is only sensitive for detecting deletions that are of the order of several megabases in size, and that smaller deletions or balanced alterations may thus be missed, the lack of DNA copy number changes in any of our adamantinomatous and papillary craniopharyngiomas is also in accordance with previously published CGH data on low grade cerebral neoplasms: pineocytomas, subependymal giant cell astrocytomas, and pilocytic astrocytomas were reported to show 0.5, 0.12, and 0.33 imbalances per tumour, respectively. In view of our data, and most other molecular data on craniopharyngiomas, one has to assume that chromosomal aberrations do not play a major role in their tumorigenesis, and the only two cytogenetically abnormal cases may have represented tissue culture artefacts.

In conclusion, our CGH data suggest that chromosomal imbalances are a rare event in primary adamantinomatous and papillary craniopharyngiomas.

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