Chromosome 6q

Deletions of chromosome 6 are a common abnormality in diverse human malignancies including astrocytic tumors, suggesting the presence of tumor suppressor genes (TSG).

Terminal deletion of chromosome 6q is a rare chromosomal abnormality associated with intellectual disabilities and various structural brain abnormalities.

Iwamoto et al. presented a case of 6q terminal deletion syndrome with unusual magnetic resonance imaging (MRI) findings in a neonate.

The neonate, who was prenatally diagnosed with dilation of both lateral ventricles, was born at 38 weeks of gestation. MRI demonstrated abnormal membranous structure continuing to the hypertrophic massa intermedia in the third ventricle that had obscured the cerebrospinal fluid pathway, causing hydrocephalus. G-band analysis revealed a terminal deletion of 6q with the karyotype 46, XY, add(6)(q25.3) or del(6)(q26). He underwent ventriculoperitoneal shunt successfully, and his head circumference has been stable.

6q terminal deletion impacts the molecular pathway, which is an essential intracellular signaling cascade inducing neurological proliferation, migration, and differentiation during neuronal development. In patients with hydrocephalus in association with hypertrophy of the massa intermedia, this chromosomal abnormality should be taken into consideration. This case may offer an insight into the hydrocephalus etiology in this rare chromosomal abnormality [1)].

In order to help identify candidate TSGs, Ichimura et al have constructed a chromosome 6 tile path microarray. The array contains 1,780 clones (778 P1-derived artificial chromosome and 1,002 bacterial artificial chromosome) that cover 98.3% of the published chromosome 6 sequences. A total of 104 adult astrocytic tumours (10 diffuse astrocytomas, 30 anaplastic astrocytomas (AA), 64 glioblastomas (GB)) were analysed using this array. Single copy number change was successfully detected and the result was in general concordant with a microsatellite analysis. The pattern of copy number change was complex with multiple interstitial deletions/gains. However, a predominance of telomeric 6q deletions was seen. Two small common and overlapping regions of deletion at 6q26 were identified. One was 1,002 kb in size and contained PACRG and QKI, while the second was 199 kb and harbours a single gene, ARID1B. The data show that the chromosome 6 tile path array is useful in mapping copy number changes with high resolution and accuracy. We confirmed the high frequency of chromosome 6 deletions in AA and GB, and identified two novel commonly deleted regions that may harbour TSGs [2)].

Rennert et al report an unusual case of a widely glioblastoma multiforme metastasis. DNA copy number microarray profile of the resected specimen revealed complex rearrangements found throughout chromosome 6, a phenomenon known as chromothripsis. Such chromothripsis pattern was not observed in 50 nonmetastatic glioblastoma specimens analyzed. Analysis of the 1000+ gliomas profiled by The Cancer Genome Atlas (TCGA) data set revealed one case of chromosome 6 chromothripsis resembling the case described here. This TCGA patient died within 6 months of

undergoing tumor resection. Implications of these findings are reviewed in the context of the current literature [3].

1)
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2)
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3)
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