Association of autism with polyomavirus infection in postmortem brains.

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Autism is a highly heritable behavioral disorder. Yet, two decades of genetic investigation have unveiled extremely few cases that can be solely explained on the basis of de novo mutations or cytogenetic abnormalities. Vertical viral transmission represents a nongenetic mechanism of disease compatible with high parent-to-offspring transmission and with low rates of disease-specific genetic abnormalities. Vertically transmitted viruses should be found more frequently in the affected tissues of autistic individuals compared to controls. Our initial step was thus to assess by nested polymerase chain reaction (PCR) and DNA sequence analysis the presence of cytomegalovirus (CMV), Epstein-Barr virus (EBV), herpes simplex virus type 1 (HSV1), herpes simplex virus type 2 (HSV2), human herpes virus 6 (HHV6), BK virus (BKV), JC virus (JCV), and simian virus 40 (SV40) in genomic DNA extracted from postmortem temporocortical tissue (Brodmann areas 41/42) belonging to 15 autistic patients and 13 controls. BKV, JCV, and SV40 combined are significantly more frequent among autistic patients compared to controls (67% versus 23%, respectively; P < .05). The majority of positives yielded archetypal sequences, whereas six patients and two controls unveiled single-base pair changes in two or more sequenced clones. No association is present with the remaining viruses, which are found in relatively few individuals (N <or= 3). Also polyviral infections tend to occur more frequently in the brains of autistic patients compared to controls (40% versus 7.7%, respectively; P = .08). Follow-up studies exploring vertical viral transmission as a possible pathogenetic mechanism in autistic disorder should focus on, but not be limited to, the role of polyomaviruses.