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Isochromosome 9p and choroid plexus papilloma: Coincidence or cause? J.M. Fischer and H.V. Toriello. Genetic Services, DeVos Women and Children's Hospital, Grand Rapids, MI.

Choroid plexus papilloma (CPP) is a generally benign brain tumor that forms within the ventricle and may lead to hydrocephalus. Two-thirds of patients with CPP are under the age of 2 years. CPP has been associated with von Hippel-Lindau syndrome, Aicardi syndrome and hypomelanosis of Ito. Non-syndromic CPP may be inherited in an autosomal recessive pattern. It has also been proposed that duplication of 9p is associated with hyperplasia of the choroid plexus. Our patients, 1 2/12 year old twin girls, presented for genetic evaluation due to suspected CPP. Both patients had height and weight below the 3rd percentile and head circumference at or just below the 50th percentile. Both patients also had deep-set small eyes, prominent nose, normal appearing mouth and ears, single palmar creases, bilateral 5th finger clinodactyly, hyperconvex nails of fingers 3, 4, and 5, spatulate fingertips, and hypoplastic toenails. One of the twins functioned at the 7-8 month level developmentally, had varus position of the foot and a large anterior fontanelle. The second twin functioned at the 13 month level developmentally with areas of concern being language delay, fine motor skills, and self-help issues. The second twin also had strabismus. Due to the multiplicity of findings, we were suspicious of a chromosomal aberration possibly involving the p arm of chromosome 9. Peripheral blood chromosome analyses using banding and multicolored FISH revealed a karyotype of 47,XX,+idic(9)(q12) or tetrasomy 9p. Review of the literature revealed few cases of tetrasomy 9p and variable expression thereof. Though hydrocephaly and Dandy-Walker malformation had been reported in some individuals, only 2 cases included abnormalities of the choroid plexus, CPP and hyperplasia respectively. Our patients appear to have a less severe expression of tetrasomy 9p. This case demonstrates that CPP may be yet another finding of tetrasomy 9p. In addition, it may further support the suggestion that a gene on 9p controls growth of the choroid plexus.

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Terminal deletion of 11q in two brothers: Clinical, cytogenetic, molecular genetic and counseling issues. M.M. Haag, S.M. Phillips, M.L. Tunby L.S. Beischel, C.L. McCann, J.C. Hansen*, J.P. Johnson, J.F. Reynolds. Montana Medical Genetics Program, Shodair Hospital, Helena, MT and *Medical Associates, P.C., Bozeman, MT.

The 11q- syndrome (Jacobsen syndrome) involves characteristic but variable features including trigonocephaly, facial dysmorphism, psychomotor retardation, cardiac defects, digital anomalies and thrombo- or pan- cytopenia. Most cases of 11q- syndrome are *de novo*, and involve terminal or interstitial deletions at 11q23 and distal. Molecular studies of deletion breakpoints have delineated a phenotypic map for some of the major features. Expression of a rare folate-sensitive cytogenetic fragile site at 11q23.3, FRA11B, has been attributed to expansion of a CCG repeat in the region. A few cases of *de novo* terminal deletions of 11q apparently result from breaks at the FRA11B site in offspring of individuals with fragility and/or CCG expansion. We report here the first case, to our knowledge, of recurrent terminal deletion of 11q in two brothers. Both boys presented at birth with thrombocytopenia and mild dysmorphic features including epicanthal folds, broad depressed nasal bridge, hypertelorism, brachycephaly, and undescended testes. The propositus, at 10 months, was referred to Genetics clinic for evaluation of developmental delay. His older brother, evaluated elsewhere, was mosaic for 11q-. Chromosome studies on the propositus and a repeat study on his older brother revealed a del(11)(q24). The older brother had normal 11's in 12% of cells at 30 months of age. *FISH* with the MLL probe at 11q23 and the subtelomeric probe for 11q showed that the breakpoint in these patients is distal to MLL, and that the telomeric probe is deleted from one homolog in all cells of the propositus and most cells of his brother with mosaicism. Parental chromosomes are normal, and FRA11B was not observed in cells grown in folate-deficient media. Molecular studies are in progress to: 1) determine the number of triplet repeats at FRA11B; 2) more precisely determine the breakpoint and parental origin of the deletion; 3) attempt to elucidate the cause for the familial recurrence and 4) counsel the family for reproductive options.

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**FISH delineation of multiple chromosome abnormalities in a mentally retarded patient with severe chronic disabilities.** K. B. Harrison,[1] G. Eddey,[2] G. Barabas,[2] J. Mintz[2]. [1]Morristown Memorial Hospital, Morristown, NJ and [2]The Matheny School and Hospital, Peapack, NJ.

We report a severely mentally retarded 17-year old girl with congenital abnormalities, seizure disorder, cerebral palsy, hydrocephalus, and numerous constitutional chromosome abnormalities.

She was born at term via emergency C-section to healthy, nonconsanguinous parents. There were no reported teratogen exposures. Chromosome studies revealed several abnormalities which could not be fully defined. Hydrocephalus was noted at two months of age and corrected with a VP shunt. Maternal and sibling chromosomes were normal. Paternal chromosomes studies were not done.

The prometaphase G-banded karyotype of the patient shows several abnormalities involving chromosomes 1, 6, 7, 8, and 10. FISH with various paints and probes have defined an apparently reciprocal translocation between chromosomes 1q and 8q, translocation of portions of 6q onto 7q and 10q, and insertion of a portion of 10q into 1q.

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**Electronic karyotype transmission**. K.B. Harrison[1] and D. Warburton[2]. [1]Department of Pathology, Morristown Memorial Hospital, Morristown, NJ and [2]Departments of Genetics and Development, and Pediatrics, Columbia University, New York, NY.

The Internet offers a wealth of resources for clinical geneticists. Literally millions of sources of specialized information, search engines, listserves, patient support resources, and online conferencing are available and enhance knowledge, efficiency, and patient care. For the clinical cytogeneticist, internet transmission of high resolution karyotypes and FISH images provides another valuable electronic tool. Pictures can be downloaded from an imaging system onto a floppy disk and e-mailed after removal of patient identifiers. They can be viewed with a simple graphics program and the results quickly reported via return email or telephone. This remote case review by the laboratory director or a designee solves the majority of laboratory coverage problems and maintains turnaround time and efficiency. In addition it provides immediate karyotypes or FISH images when consultation is needed. Emailing of images allows sharing of data among colleagues regardless of differences in computerized image
will require microsco
the few drawbacks o