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Distal 5q Trisomy resulting from an X;5 translocation detected by chromosome painting. D. N. Abuelo[1,3], A. N. Ahsanuddin[1] and H. F. L. Mark[1,2]. [1]Brown University School of Medicine, [2]Lifespan Academic Medical Center Cytogenetics Laboratory and [3]Genetic Counseling Center, Rhode Island Hospital, Providence, RI.

The distal 5q trisomy genotype has been associated with clinical signs including short stature; microcephaly; downward palpebral slant, strabismus; prominent, widened nasal bridge; long, flat philtrum; long, thin upper lip with downturned corners; large, low-set, dysplastic ears; limb and joint malformations; cardiopulmonary abnormalities; abdominal hernias; and mental retardation. We describe a 13-year-old female with an apparently *de novo* unbalanced translocation resulting in the presence of additional chromosomal material on the short arm of the X chromosome, detected by conventional G-banding studies. She exhibits several of these features: short stature, prominent nasal bridge, flat philtrum, thin upper lip, limb and cardiac malformations. Fluorescent *in situ* hybridization (FISH) using the Chromoprobe™ Multiprobe-M protocol demonstrates that the additional chromosomal material originates from chromosome 5. The karyotype of this patient is now established to be 46,X,der(X)t(X;5)(p22.3;q33). This is the first case of distal 5q trisomy arising from a translocation with the X chromosome.

Replication studies on this patient show that the derivative t(X;5) chromosome is late replicating in almost all cells examined, which indicates that this chromosome is preferentially inactivated. However, the translocated segment of chromosome 5 appears to be early replicating, which implies that the trisomic 5q segment is transcriptionally active. We cannot determine from these studies whether all or only some genes in this segment are expressed, but this patient's relatively mild clinical signs suggest that the critical region(s) which contribute to the distal 5q trisomy phenotype are at least partially suppressed.

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Trisomy 8 in papillary serous carcinoma of the ovary studied by FISH. A. Afify, M. Samy, C.-L. Sun and H.F.L. Mark. Lifespan Academic Medical Center Cytogenetics Laboratory, Rhode Island Hospital and Brown University School of Medicine, Providence, Rhode Island; University of Michigan Medical Center, Ann Arbor, Michigan.

Ovarian cancer is the leading cause of death from gynecologic malignancy among women in the United States. In 1997, there were nearly 27,000 ovarian cancer cases with over 14,000 deaths. Recent attempts at early detection of ovarian cancer have been aimed at the identification of biomarkers that would indicate an underlining malignant process or reflect the biological behavior of the tumor. Our previous studies revealed that chromosome 8 copy number abnormality, especially trisomy, is common in several cancers. Archival tissues from 24 cases of papillary serous ovarian carcinoma (10 stage I and 14 stage III) were analyzed by FISH with a chromsome 8 α-satellite probe (Oncor, Gaithersburg, MD). The analysis was done according to standard protocols of the Lifespan Academic Medical Center Cytogenetics Laboratory at Rhode Island Hospital. Twenty-one out of 24 cases (87.5%) were found to be trisomic for chromosome 8, if a cutoff point of ≥15% cells with three signals is adopted. Overall 80% of stage I and 93% of stage III tumors had trisomy 8. This study confirms the presence of a high frequency of trisomy 8 in both early and late stages of the disease and suggests that trisomy 8 may be an early event in the multi-step process leading to ovarian cancer. (This study was funded in part by Vysis, Inc., Downers Grove, IL.)

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Chromosome painting for diagnosing a 10;11 translocation in a patient with infantile acute lymphoblastic leukemia. D. Alter, L. Glasser and H.F.L. Mark. Lifespan Academic Medical Center Cytogenetics Laboratory, Rhode Island Hospital and Brown University School of Medicine, Providence, RI.

Numerical and structural chromosomal abnormalities occur in up to 90% of cases of childhood ALL. Two-thirds of these abnormalities are recurrent. The most common abnormalities are pseudodiploidy and t(1;19), occurring in 40% and 5-6%, respectively. Hyperdiploidy has the best prognosis, with 80-90% five-year survival. The 4;11 translocation has the worst prognosis, with a 10-35% five-year survival. We report a patient with infantile acute lymphoblastic leukemia and a nonrecurrent translocation, t(10;11). Structural rearrangements between chromosomes 10 and 11 have been observed in 0.5% of all cases of childhood ALL with cytogenetic abnormalities. The identification of this apparently unique structural abnormality was achieved using fluorescent *in situ* hybridization (FISH) with chromosome 10- and chromosome 11-specific painting probes as an adjunct to conventional cytogenetics. As is often the case, suboptimal preparations often preclude unequivocal identification of complex rearrangements by banding techniques. The cytogenetic diagnosis of our patient was established as 46,XY,t(10;11)(p15q25;q14p11). The benefits of FISH serve to increase the resolution of detection for chromosomal abnormalities and the understanding of the pathogenic mechanisms of childhood ALL.

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Hypotonia and Prader Willi syndrome in the neonatal period. D. Bérubé and R.Gagné. Centre Hospitalier Universitaire de Québec, Pavillon CHUL, Québec, Canada.

Hypotonia is sometimes a diagnosis for which infants are referred for investigation in the neonatal period.. Hypotonia occurs in infants with different disorders: neuromuscular, metabolic and chromosomal disorders. Among chromosomal disorders, hypotonia is a differential diagnosis particularly in Down and Prader Willi syndromes. Thus, karyotyping and fluorescent *in situ* hybridyzation with specific probes should be considerd if a child is hypotonic and/or dysmorphic. In our laboratory, since almost two years, we have begun a study in neonates and children under six months referred for hypotonia. The aim of this study is to detect very early in infancy children with Prader Willi syndrome. For each case, routine chromosome analysis is performed. In all cases. When no cytogenetic chromosomal abnormalities have been found, a fluorescent *in situ* hybridyzation technique with SNRPN probe has been performed. The SNRPN probe detect 75 % of deletion in the Prader Willi syndrome region, the 25% of undected cases needed more investigation by other molecular techniques like research of maternal uniparental disomy The results showed that in cases referred only for hypotonia, around 15 % of cases have a microdeletion on chromosome 15 q11.2. The results showed a significative detection rate for pursuing the use of fluorescent *in situ* hybridyzation in cases of hypotonia in neonates and children under six months of age. The early detection in Prader Willi syndrome is very important because children with Prader Willi syndrome develop a morbid obesity and complication resultin from this obesity. Management is aimed at prevention of obesity and treatment of its consequence.