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Mosaicism for duplication of 17q21 ·qter with lymphedema and normal phenotype. M. Descartes, L. Baldwin, P. Cosper, A. Carroll. Department of Human Genetics, University of Alabama at Birmingham, Alabama.

Duplication of 17q21 ·qter is associated with a clinically recognizable syndrome. The major features are: profound mental retardation; dwarfism; frontal bossing and temporal retraction; narrowing of the eyes; thin lips with overlapping of the lower lip by the upper lip; abnormal ears; cleft palate[1]. The region that appears to be responsible for the phenotype is 17q23 ·qter[2]. Serothken et al. reported an infant mosaic for the duplication 17q21.1 ·qter, their patient had many features suggestive of the 17q duplications syndrome except for the craniofacial dysmorphism[3]. We report an infant who was found to be mosaic for duplication 17q21 ·qter who had none of the features associated with this syndrome.
Our patient was the second child of a 24 year old mother and a 28 year old father. The pregnancy was complicated with an abnormal prenatal ultrasound that showed mild-moderate fetal ascites and mild pericardial effusion at 24 weeks gestation. Genetic amniocentesis for karyotype analysis showed: 46,XX,dup(17)(q21q25)[8]/ 46,XX[12]. Parental chromosomes were both normal. The patient was first seen by genetics at age one month. Karyotype analysis in leukocytes was done and reported to be normal. Chromosome analysis of cells derived from skin was done 4 months later after the patient was noted to have swelling of her extremities. The fibroblast chromosome analysis came back abnormal: 46,XX,dup(17)(q21q25)[4]/46,XX[16]. At twelve months of age the patient's weight, length and head circumference were on the 50th percentile with normal development. On examination she had: frontal bossing; narrow palpebral fissures; flat nasal bridge; epicanthal folds; short nose; pointed chin; short neck; toe nail hypoplasia. Some of the above described facial features were also present in her parents. The family history was negative for lymphedema. Our patient is the first reported case of dup 17q and dup 17q mosaicism with lymphedema and essentially normal phenotype.

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Trisomy 8 in cervical cancer. D. Feldman, S. Das, H. Kye, C.-L. Sun, M. Samy and H. F. L. Mark. Lifespan Academic Medical Center Cytogenetics Laboratory, Rhode Island Hospital and Brown University School of Medicine, Providence, RI.

Cervical cancer is a malignancy which typically occurs at the transformation zone between squamous and glandular epithelium. The vast majority fall into two histologic types: squamous cell and adenocarcinoma. We have previously reported extensively on abnormal chromosome 8 copy number in various cancers, which appears to be an ubiquitous phenomenon. In the present pilot project, we studied chromosome 8 copy number together with a chromosome 17 control using formalin-fixed paraffin-embedded archival cervical cancer tissues. HER-2/neu oncogene amplification was also studied in this sample, as reported in a previous abstract presented at the 1998 Annual Meeting of the American Society of Human Genetics. From a total of 23 cases successfully analyzed, 12 (52%) were found to be trisomic for chromosome 8, and 3 (13%) were found to be trisomic for chromosome 17. Of the trisomic 8 cases, 2 were also trisomic for chromosome 17, which implies that the tumors are either triploid or doubly trisomic. However 10 tumors were trisomic for chromosome 8, but were disomic for chromosome 17. Among the 12 trisomic 8 cases, 1 was also amplified for HER-2/neu whereas 11 were nonamplified. Clinicopathologic parameters of the trisomic and amplified cases will be compared to those of the disomic nonamplified cases. In view of the present results, further exploration using a larger study sample size may be warranted. (This study was partially funded by Vysis, Inc., Downers Grove, IL).

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A Dermatofibrosarcoma Protuberans (DFSP) with complex clonal chromosomal findings and absence of ring chromosomes: G.Hostetter, J.Freeman, R.Naeem, Baystate Medical Center, Springfield MA and Tuft Medical School Boston.

Recent studies have characterized dermatofibrosarcoma protuberans (DFSP) with ring chromosomes consisting of interspersed sequences from chromosome 17 and 22. We present a case of a classical DFSP with complex clonal chromosomal aberrations and normal chromosome 17 and 22 with no ring chromosome. A 29 year old male presented with back lesion. Histologic features included a monotonous population of spindle cells in storiform pattern. The mitotic activity was relatively high but the cells lacked nuclear atypia and necrosis. Immunohistochemical staining was strongly positively for CD34 in a membranous pattern of all tumor cells. Outside expert consultation confirmed the diagnosis of DFSP.
Cytogenetic evaluation from G banding and FISH revealed a clonal complex male karyotype with a consistent finding of an additional chromosome material of unknown origin on chromosome 19 at band 13 and no ring chromosome. Interestingly this der 19 chromosome finding has been reported as a feature in 25% of malignant fibrous histiocytomas with tumor progression which has some similarity with DFSP. This case either represent a limitations of classical histopathology or similar mechanism of origin of these two tumor types. It may also represent tumor progression and dedifferentiation with loss of ring chromosome.

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Molecular Cytogenetic Characterization of Chromosome Markers. CB Lozzio, L Lyall, and E Bamberger Developmental and Genetic Center, Univ of TN Medical Center, Knoxville

The clinical significance of marker chromosome depends on the origin of the marker, size and presence of euchromatin. We report the results of fluorescence "in situ" hybridization (FISH) studies on ten marker chromosomes. Six of these markers were derived from chromosome 15 and ranged in size from a small bi-satellite supernumerary marker to a large isodicentric inverted duplication including the euchromatic region 15q11-q13. This large marker was found in a 25-year-old severely mentally retarded male with oval face, seizures and abnormal speech and gait. Two of the medium size inv dup(15) were detected prenatally. These markers had a small euchromatic region labeled with the whole chromosome 15 probe and no signal for GABR3 and SNRPN of the 15q11-q13 region. Two chromosome 18 markers were analyzed and one of them was an isochromosome 18p labeled with 18p specific probe. The other two markers were derived from chromosome 13. One was a small centromeric supernumerary chromosome, the other was a small metacentric chromosome with two signals for the 13q32-q33 region and for the telomeric probe 13q34→qter and no signal for any alpha satellite centromere. Thus, this isodicentric 13q marker had a functional neo-centromere. The patient was a mosaic with one and two extra markers causing tetrasomy and polysomy 13q32→qter and clinical features of trisomy 13q3.