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Duplication 17p11.2 in two patients: clinical features and molecular cytogenetic findings. C.M. Powell, A.S. Aylsworth, K.A. Kaiser-Rogers, K.W. Rao. Univ. of North Carolina, Chapel Hill, NC.

Since patients with duplications or deletions involving the Smith-Magenis syndrome (SMS) region at 17p11.2 are thought to arise by the same mechanism of unequal recombination between a repeat sequence that flanks the SMS region (Chen et al., 1997), it is surprising that the duplication patients have been so rarely reported (Magenis et al., 1986; Kozma et al., 1991; Brown et al., 1996; Chen et al., 1997 and 1998; Summers et al., 1998). These patients are most likely underdiagnosed because of lack of defined clinical features and subtlety of the duplication with routine cytogenetic studies. We report two additional patients and review the common physical features in this duplication syndrome.

Patient 1 is an 11 month old male with postnatal growth retardation, length at mean for 5 months, and head circumference average for 6 months. He has ptosis of the left eyelid, narrow nose and palate, small mandible, sacral dimple, overlapping toes, decreased tone, and global developmental delay. Patient 2 is a 6 year old female with a history of failure to thrive in early childhood and developmental delay. Height, weight, and head circumference are at the 50th percentile; she has upslanting, narrow palpebral fissures, broad eyebrows, distal joint laxity, and normal neurologic exam. Developmental testing at age 6 showed a full scale IQ of 65, with performance IQ 60 and verbal IQ 78 and behavior consistent with attention deficit hyperactivity disorder.

Cytogenetic analyses with G-banding showed duplication of 17p11.2 with a narrow extra gray band in the middle of the G-negative area in the p11.2 region. The duplication was confirmed with a FISH assay using the D17S258 probe (Oncor) for the SMS region. The D17S122 cosmid probe (Oncor), which hybridizes within the region that is duplicated in Charcot-Marie-Tooth type 1A patients, showed a single signal on each chromosome 17 indicating that this region is not duplicated.

Comparison of facial features reveals striking similarities between patient 2 and the patient reported by Magenis. It also appears that despite significant early problems with weight gain and development, these patients have fewer problems as they get older and may function in the low normal to borderline range. Detailed clinical descriptions of patients with duplications of 17p11.2 will be helpful in further delineation and recognition of this duplication syndrome.

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22q13 Deletion Syndrome: A Genetic Basis For Neurobehavioral Disorders? C Prasad[1], B N Chodirker[2], C Lee[3], A K. Dawson[4], L J Jocelyn[5], and A E Chudley[2] [1]Medical Genetics, Memorial University, St. John's Newfoundland A1B3V6, [2] Section of Genetics and Metabolism, Department of Pediatrics and Child Health, [4]Cytogenetics, Health Sciences Centre, [5]Child Development Clinic, University of Manitoba, Winnipeg, MB R3EOZ2, [3]Department of Pathology and Laboratory Medicine, IWK Grace Health Centre, Halifax, Nova Scotia B313G9.

Neurobehavioral disorders pose a challenge to physicians, both in diagnostic and therapeutic aspects. Many neurobehavioral disorders have been identified to have a genetic basis such as the fragile X syndrome and the 22q11 deletion associated with the Velocardiofacial syndrome. We describe three children with a different and less well characterized deletion involving the most distal band of chromosome 22 (22q13), where the presentation entails deviant development, minor dysmorphism and autistic features.

All 3 patients had developmental delay and unusual behavioral features. All our patients exhibited normal to advanced growth. Patient 1 had an added feature in submucous clefting of the palate. Patient 2 and 3 shared features of autistic spectrum disorder. Results of cytogenetic analysis in the three patients are as follows: Patient 1 (46XX ish del (22) (q13.3) (D22S39- D22S75X2), Patient 2 (46XY, del(22)(q13.1), Patient 3 (ish del (22)(q13q13) (LSI ARSA-) de novo). Thus, all three patients appear to share a deletion affecting the terminal 22q13 region.

Literature review of the patients with 22q13 deletion is presented. Initially, the deletion was detected using high resolution chromosome analysis. Recently a FISH probe (D22S39) through its fortuitous use as a control probe in testing for 22q11 deletions has been used to demonstrate this deletion. The phenotypic features in most of the cases described include marked expressive speech delay, deviant development and pervasive behaviors, minor motor delays, minor dysmorphism, with normal or exaggerated growth. While there remain gaps in our understanding of this particular deletion syndrome, we propose that patients with significantly delayed speech, deviant development and behavioral issues, and minor facial dysmorphism be screened for this deletion.

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A multifaceted genetics educational program for nursing faculty. C.A. Prows[1], K.K. Latta[1] & C. Hetteberg.[2] [1]Children's Hospital Medical Center, Cincinnati, OH, [2]Univ. of Cincinnati, OH.

The 1998 *Statement on the Scope and Standards of Genetics Clinical Nursing Practice* asserts that all nurses should be able to identify, refer, support and care for persons affected by, or at risk for manifesting or transmitting, genetic conditions. Nurses also need to play a significant role in the management of genetic information. However, results from multiple surveys conducted over the past 26 years have repeatedly demonstrated that nurses receive limited genetics education in their basic nursing programs. Nursing faculty's limited educational preparation in genetics is a significant barrier to increasing genetics content in nursing curriculum. The *Summer Genetics Program for Nursing Faculty* was developed to address the genetics continuing educational needs of nursing faculty. One component of the program was a 12-day Genetics Summer Institute (SI). In 1997, 26 nursing faculty attended the first SI and in 1998, 34 additional nursing faculty attended the second SI. These 60 participants represented 59 nursing schools from 25 states and the District of Columbia. Change in participants' genetics knowledge was measured using a 35 item pre / post-test. Both participant groups demonstrated statistically significant improvement ($p<0.01$) in post-test scores. Follow up of 1997 participants has occurred twice since the first SI to assess participants' progress toward increasing genetics content in their curricula. Results indicate that 75% used information and educational resources provided during and following the SI in their own and/or fellow faculty's existing lectures. Nearly all discussed the importance of genetics with their school administrators and fellow faculty. Over 80% pursued further genetics continuing education. Seven of the 1997 participants developed a genetics course. Twice yearly follow-up is underway for the 1998 participants and will continue for the 1997 participants. This program was funded by the NHGRI's Ethical, Legal and Social Implications Research Program at NIH.

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Heart-hand syndrome II (Tabatznik syndrome): A new case with mild phenotype. F.J. Ramos[1], I.Bueno[1], J.L. Olivares[2], Bueno M[2]. [1]Secc. Genetica, [2]Dept. Pediatria, Hospital Clinico Universitario, Fac. de Medicina Univ de Zaragoza.SPAIN

In 1978, Temtamy and mcKusick reported a family observed by Tabatznik in which the affected patients had upper limb malformations and cardiac arrhytmia. Dominant either autosomal or X-linked, inheritance was proposed. Hand malformations included brachytelephalangy of the thumb and mild shortening of the IV and V metacarpals. Phenotypically, all the affected individuals had, among other features, sloping shoulders and short upper limbs.

After that first report, Lala et al., in 1990, published a family in which the 8-year-old proband had similar cardiac and upper limb anomalies than the cases of Tabatznik, in addition to another features such as facial dysmorphism, cryptorchidism and mild mental retardation.

In 1980, Ruiz de la Fuente and Prieto reported on what they called a new type of autosomal dominant heart-hand syndrome (type III). In that form, affected patients had type C brachydactyly associated with cardiac arrhytmias. No upper limb anomalies, beside those of hands and feet, were observed. The cardiac arrhytmias consisted of intraventricular conduction defects and sick sinus syndrome.

Here, we present a new case of Heart-Hand syndrome type II in a 8-year-old girl who had bilateral IV and V short metacarpals and metatarsals, and cardiac arrhytmia consisting in abnormal migration of sinusal pacemaker. She also had a mild mitral and tricuspid prolapse without clinical symptoms. She had some facial dysmorphic features and normal intelligence. Family history showed the presence of hand anomalies in some individuals of the maternal side.

Differential diagnosis with other Heart-Hand syndrome types will be discussed.