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Fetal Valproate Syndrome (FVS) through the USF Teratogen Information Service (TIS). B.G.Kousseff, S. Sage and G. Jervis, Division of Medical Genetics, University of South Florida, Tampa, Florida.

Between January 2, 1988 and December 31, 1997 through the TIS program and genetics clinics at USF there were 168 requests for information/evaluations about valproic acid exposure. Fifty-four of these were for valproic acid as monotherapy. Among them, 7 were for information only, 3 women were not pregnant, 3 have not delivered yet and 2 were for paternal exposure. Of the remaining 39 pregnancies, there is no outcome information on 19. Of the 20 referrals with reported outcome, there are 13 full term normal infants, 1 premature without abnormalities, 1 hydropic premature stillborn and 5 with anomalies: 1 case each of hypoplastic left heart, hypospadias, club feet, single umbilical artery and anisocoria. Among the 114 referrals for combination therapy there were 6 pregnancies with fetal anomalies; 2/6 had open neural tube defect (ONTD) with hydrocephaly and one of them had polysyndactyly as well. The latter had normal karyotype and the mother took 750 mg valproic acid/day and paroxetine 25 mg/day. The other mother was on valproic acid, haloperidol and chlorpromazine (dosage unknown) for chronic schizophrenia with seizures. The third abnormal outcome was a term boy with trigonocephaly, right hand preaxial polydactyly, right inguinal hernia and pyloric stenosis. The mother took valproic acid 2500 mg/day and Phenobarbital 120 mg/day for seizures since infancy. The remaining three infants had VSD, polydactyly and sacral teratoma respectively. These data support the valproate teratogenicity in humans by showing ONTD in 2/168, polysyndactyly in 3, congenital heart defects in 2, inguinal hernia in 1 and hypospadias in 1. Trigonocephaly, pyloric stenosis, hypoplastic left heart, anisocoria and sacral teratoma suggest an expanded FVS phenotype. In 1 of the 2 infants with ONTD the mother had seizures due to MVA so epilepsy was excluded as a possible teratogen. Also the combination therapy in both mothers did not consist of two anticonvulsants; valproic acid was combined with paroxetine in one and with haloperidol and chlorpromazine in the other. As to TIS as an adjunct in addressing/recording/managing birth defects, important outcome information can be lost without an active follow-up component. However, a TIS program is invaluable for obtaining data regarding prenatal exposures.

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45,X/46,XY Mosaicism: Clinical management and Counseling Issues

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Individuals with 45,X/ 46,XY mosaicism can present variant phenotypes ranging from normal appearing males to females with the clinical manifestations of Turner Syndrome. In males, hypospadias, undescended testis, and Muellerian remnants are quite common.
In all individuals with 45X/46,XY, mosaicism, the risk of gonadal malignancies is increased. In order to prevent complications associated with gonadal dysgenesis we recommend routine testicular biopsies in newly diagnosed male patients, and routine follow up with pediatric urologists in all patients. We report two new cases of 45,X/46,XY and discuss the clinical and counselling issues involved.

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Genetics education of health professionals, E.V. Lapham[1], J.O. Weiss[2], C. Kozma[1], J.L. Benkendorf[1], M.A. Wilson[2] [1]Georgetown Univ., DC, [2]Alliance of Genetic Support Groups, DC.

Emerging genetic advances mandate that all health professionals have basic knowledge of genetics in order to provide adequate services to their clients. As part of the Human Genome Education Model (HuGEM) Project II, a national survey was carried out by the survey Research Center, U. MD., of the genetics education, perceived competencies, and views of ethical issues of nearly two thousand health professionals (audiologists, dietitians, occupational therapists, physical therapists, psychologists, social workers, and speech-language pathologists). Completed in September, 1998, the survey showed that most health professionals are providing some genetic services to their clients. Overall, 70% have discussed the genetic component of their clients' problems and 30% have provided counseling about genetic concerns with at least a few of their clients. Yet only 17% have high or moderately high confidence in their ability to discuss genetic basis of clients' genetic conditions and 14% are confident in providing guidance to clients with genetic conditions. Fifty-six percent take medical histories, however, only 23% feel confident in their ability to elicit genetic information as part of the medical history. While 67% of health professionals hold graduate degrees, fewer than 21% have had one or more courses in genetics. Within the last 6 months, 83% have received genetics information from TV/Radio, 32% from conferences, and 28% from clients. Nearly two-thirds would like to have continuing education in genetics. The top priority topics were 1) Role of genetics in common disorders such as stroke, heart disease, and cancers, 2) Overview of human genetics, 3) Genetic information and racial/ethnic concerns, 4) Helping clients cope with a new genetic diagnosis, and 5) Identifying genetic resources. These data imply that most health professionals are already providing genetic services to at least some clients, few have high confidence in providing the services, and most have had little or no education in genetics. Most respondents want continuing education in genetics and identify priority topics for education. This information has significant implications for genetics educators.

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Pregnancy Termination Policies in Community Hospitals. R.R. Lebel(1,2), G.A. Lofthouse(1).
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Many community hospitals with religious historical foundations hold strict policies against pregnancy termination. Prenatal detection of severe malformations and/or genetic disorders in the fetus provides opportunity for choices regarding the continuation or termination of pregnancy, posing quandaries. Patients often must travel significant distances and meet entirely new health care providers at a juncture when emotion and anxiety levels are high. We present a policy, in a religiously based community hospital, designed to allow termination of pregnancies in which lethal conditions have been detected by prenatal testing. We describe the process by which a multidisciplinary committee worked through ethical, moral, social and religious issues to craft a document which was perceived as acceptable within the context of the hospital tradition while also meeting patient needs.