platform presentations in clinical genetics

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A retrospective review of neurobehavioral and psychosocial issues in adults with putative Sotos syndrome. R.R. Anderson and G.B. Schaefer. Univ. Nebraska Medical Center, Omaha, NE.

Families in the Sotos Syndrome Support Association with affected individuals aged 21 or older were invited to participate in a telephone interview. All families with current phone numbers assented. In two cases, the interview was with the affected individual; the balance with a parent. Medical status, educational history, employment experience, living arrangements, life skills, goals and aspirations, happiness and spiritual life were explored during the telephonic interview. Medical records and photographs were requested thereafter to provide a more complete assessment, and review the reliability of the Sotos diagnosis. Of 16 probands ages 20 - 35, 10 live with their parents; 4 in group homes; 1 at college and 1 in assisted apartment living. 2 are in college and 2 hold college diplomas. 13 of 16 work at least part-time, 5 in "open market" jobs and 8 in supported employment; two are considered unable to hold a job. Math skills are described as very poor, and money management is impossible for most. 5 of 16 have significant psychiatric disturbance including incapacitating anxiety, depression, aggressiveness, OCD, and schizo-affective disorder. A variety of somatic complaints have presented in adulthood, with obesity being a significant difficulty for many females. Most probands have fewer friends and outside activities than their parents would like. Few are dating or exhibiting any desire to do so. Many read for pleasure, correspond by computer, enjoy videos and music, and maintain collections of various sorts. Most are affiliated with a community of faith and are described as reasonably happy, the exceptions being those with psychiatric disturbance. The SSSA provides information and support to families of individuals with Sotos and related conditions. Adult probands may have received their diagnoses before systematic criteria were developed. Diagnostic difficulties and inherent bias of ascertainment complicate interpretation of the data gathered from support groups.

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Comparing satisfaction with clinical genetic services to other health services using a standardized survey, the CSQ-8© D.B.Flannery[1], S.T.Kozel[1], J.L.Waller[1], B.M.Ramage[2], and G.Pullen[3] [1] Med. Coll of GA, Augusta, [2] Greenwood Genetic Center, SC, and [3] Tulane Univ., New Orleans

We report the first large scale study of client satisfaction with clinical genetic services, employing a widely-used instrument, the Client Satisfaction Questionnaire (CSQ-8©) which allowed comparison of satisfaction with genetic services and published reports of satisfaction with a wide variety of other healthcare services. The CSQ-8© is a self-administered global measure of client satisfaction using 8 questions which are scored from 1 to 4, with higher scores indicating greater satisfaction. The CSQ-8© is scored by summing the individual items, producing a total score range of 8 to 32. This instrument has been used in a large number of studies, has well-established norms, excellent internal consistency, reliability and validity. The Southeastern Regional Genetics Group conducted a satisfaction survey of patients at multiple clinical genetics centers in the Southeast as part of a quality assurance program. A total of 1100 CSQ-8© forms were distributed to 10 participating clinical genetic centers. Over a 4 week period every patient seen, or their parents, were instructed to complete the anonymous survey by clinic staff after their Genetics visit. 455 surveys were returned. All 8 questions were completed on 449 of these forms, which were then used in the data analysis. The mean total satisfaction score for genetic clients was 30.15, (SD 2.66). This mean total score is higher than that of any published study of CSQ-8© scores in healthcare or counseling services. Two sample t-tests were used to determine if differences in the mean CSQ-8© scores existed between the current study and published reports of CSQ-8© scores from other healthcare services. All published studies analyzed had significantly lower mean CSQ-8© scores than this study, demonstrating that the genetic clients had better satisfaction. P-values ranged from 0.026 to less than 0.00001. High client satisfaction with genetic services should interest insurers and managed care contractors. The high satisfaction scores are particularly striking when one considers that the majority of clinical genetic encounters are new patient encounters, rather than patients involved in continuous care with the genetics team. Further analysis of this dataset will determine if satisfaction differed between pre-natal and non-prenatal visits. Future studies anticipated include testing hypotheses of causes of the high satisfaction, and assessing the use of CSQ-8 sub-item scores as a benchmarking tool between genetic centers.

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Trends in a clinical genetics program for adults. QK Gordon, HEC Hixon, MT Scheuner, Cedars-Sinai Medical Center, Los Angeles, CA.

The purpose of this study was to review trends in physician referrals, indications for referral, and payor mix for genetic consultations for common, chronic conditions of adulthood. There were 57 patient visits in 1997, 126 in 1998, and 157 in the first 3 quarters of 1999. The gender and age distribution did not vary from year to year. In 1999, 90% of patients were female, and their average age was 51 (range 22-77). 65% of patients were referred by a physician in 1998, and this proportion increased to 80% in 1999, p=0.006. The majority of patients were referred because of a family or personal history of breast or ovarian cancer; this accounted for 65% of consultations in 1998 and 71% in 1999. In these years, only 20% and 11% of patients were referred because of a personal or family history of colon cancer/polyposis, respectively. Histories of other cancer types accounted for 8% of referrals in both years. Only 4 patients (3%) were referred for cardiovascular risk assessment in 1998 and 10 (6%) in 1999. For both years, 3 patients were referred for hemochromatosis and 2 for porphyria. In 1998, 20% of patients paid cash for their genetic consultation, 20% were MediCal/Medicare, and the remainder used other forms of insurance. In 1999, only 10% paid cash (different from 1998, p=0.03), 17% were MediCal/Medicare, and 73% used other insurance. Follow-up surveys have been mailed to patients seen in 1998 with a possible inherited cancer susceptibility; thus far 36 have responded and none have reported experiencing insurance discrimination. In conclusion, these results demonstrate an exponential growth in patient referrals over the past 3 years with a significant increase in physician referrals. The largest number of patients seen have been women at risk for breast and/or ovarian cancer with disappointing patterns of referral for other cancers, cardiovascular disease and other common conditions. This trend may be due in large part to the high visibility regarding the availability of BRCA testing. The trend in increasing use of insurance for genetic consultations suggests that patients have become less fearful of insurance discrimination, which may be due to the lack of evidence that such discrimination exists.

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Is Assortative Mating the Cause for the High Frequency of Connexin 26 (Cx 26) Deafness? W.E. Nance, X-Z. Liu and A. Pandya. Virginia Commonwealth University, Richmond, VA.

Recessive mutations at Cx 26 are known to be the cause of nearly half of all genetic deafness in many populations. Since assortative mating is known to increase the variance of continuous traits and the frequency of qualitative genetic phenotypes (such as recessive deafness), it seems possible that the high frequency of Cx 26 deafness may be related to the mating structure of the deaf population. During the past 200 years, improvements in educational, social and economic circumstances have lead to an increase in the fertility of the deaf along with the appearance of assortative mating in many western populations including the United States. The new deaf by deaf mating pool has increased the frequency of common forms of recessive deafness such as Cx 26 because the non-complementary matings which produce many of the deaf offspring from these marriages are proportional to the fourth power of the gene frequency. Existing data on the frequency of non-complementary marriages among the deaf in the 19th century suggest that the incidence of Cx 26 deafness has increased in the past 100 years along with the proportion of deaf children born to deaf parents. Current observations also suggest that the high frequency of Cx 26 deafness may be confined to populations with a long tradition of intermarriage among the deaf. In Japan, for example, Fuse et al (Neuro report 10:1853,1999) found Cx 26 deafness in only four of 20 multiplex sibships (20%). The comparable rate for multiplex probands in the U.S. is 49% (Green et al, JAMA 281:2211,1999). Although we are not aware of data on the frequency of deaf by deaf matings in Japan, they were virtually unheard of in India in the past, and accounted for only about 1% of the marriages of the deaf in a reported survey from China (Liu et al, Chinese Med Genet 5:193,1988). All of these findings are consistent with the hypothesis that variation in the mating structure may have contributed to differences in the current distribution of this trait throughout the world.