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Prenatal genetic counseling by telemedicine: a feasibility study. L.A. Flore[1], S.T. Risinger[2], D.W. Britt[2], I.E. Zador[1], A.D. Gilbert[1], M.I. Evans[1] and A. Johnson.[1]
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The purpose of this pilot study was to evaluate the feasibility, technical performance, and effectiveness of prenatal genetic counseling in advanced maternal age (AMA) patients through the use of interactive video-conferencing (IVC). Eight volunteers were asked to complete a genetic history questionnaire prior to the IVC consultation. Participants received information regarding the risk of fetal aneuploidy and testing options to evaluate these risks. Counseling was provided by board certified genetic counselors. The counselor and the participants were located in separate rooms within the institution, having contact only through IVC. At each site, PC computers were networked to simulate an Internet-like connection. Additional hardware included Intel's ProShare Video System 500 with video cameras and headsets. Microsoft's NetMeeting was used for real-time video-conferencing and also for sharing custom-designed visual aid tools. Following each IVC consultation, participants were given questionnaires to assess: 1) the genetic and medical information gained during the consultation, and 2) the patient/couple's opinion regarding the efficacy of IVC genetic counseling. A protocol was developed to optimize the physical context of the system, by altering camera placement to approximate direct eye contact, and verbally orienting the patient to the unique features of the IVC equipment. All participants indicated that they believed that the counselor was able to address their needs in the IVC session. Similarly, 7/8 patients "agreed" or "strongly agreed" that they were satisfied with the consultation via IVC; 8/8 participants felt that they were "well informed" about risks and options after the consultation; all agreed that the graphic visual aids used on the IVC monitor by the counselor aided in their understanding of their genetic risks. In regard to the equipment, 3/8 indicated that the IVC made them feel uncomfortable, while 5/8 participants agreed that "it did not take long" to feel comfortable with the system. Finally, when asked if they would prefer have genetic counseling at home through IVC over standard "face-to-face" counseling, 7/8 marked "agree" or "strongly agree". These results suggest that the use of IVC for genetic counseling is a feasible alternative to traditional face-to-face consultations. To further evaluate these findings, a prospective trial in AMA patients is underway in our institution.

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Practical use of Three-Dimensional Imaging in Genetic Counseling: Patient Perception of Usefulness as a counseling tool. Jackson, DN MD, Brown L, MS, CGC, Keel-Thompson, K, RDMS. Fetal Diagnostic and Imaging Center, St. Vincent's Health Center, Department of Maternal Fetal Medicine and Fetal Imaging, Billings, MT

Aim: To assess the perceived benefit of 3-Dimensional ultrasound as an additional imaging tool for patients evaluated for abnormal ultrasound findings. Method: Aloka ultrasound system with volume mode surface rendering was offered to patients receiving evaluation at a referral maternal-fetal medicine center. Two-dimensional and three-dimensional images were obtained on all patients. Approximately 4 seconds were required for the three dimensional images to be available for patient and physician review. A follow up survey (by questionnaire) was utilized to determine patients' understanding of their specific indication. Results: Questionnaires were sent to 240 patients with abnormal ultrasound indications including echogenic bowel, renal dysgenesis, echogenic foci, cardiac structural defects, ventral wall defects, cystic hygroma, hydrops, chromosome abnormalities, neural tube defects, choroid plexus cysts, clubfeet and uterine abnormalities. Most patients (90%) strongly agreed that the information received from the 3D exam was beneficial to the visit. Parents also felt that the 3D ultrasound enhanced understanding with a recognizeable image of their baby. This finding was apparent even in pregnancies with severe or lethal outcomes. Conclusion: Ultrasound exams carry both emotional and psychological implications. The new modality of 3-dimensional scanning seems helpful in demonstrating the extent of a specific defect in a way patients understand and appreciate. Our study suggests that 3-dimensional imaging may assist in the standard evaluation of patients requiring genetic consultation for abnormal ultrasound findings.

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Prenatal diagnosis of fragile X syndrome: identification of a male fetus mosaic for a premutation on chorionic villus sampling - management and follow-up. S. J. Kennedy[1], C. Wei[2], L. Steele[2], and A. S. Teebi[1]. The Hospital for Sick Children, [1]Division of Clinical and Metabolic Genetics, [2]Department of Pediatric Laboratory Medicine, Toronto, Ontario, Canada.

Families at risk to have a child with fragile X syndrome are routinely counselled regarding the limitations of performing prenatal analysis via chorionic villus sampling (CVS) due to incomplete methylation status at this point in gestation. However, this testing option appeals to families due to the timing of the prenatal procedure. Here we report on the identification of a male fetus, by polymerase chain reaction (PCR) and Southern blot analysis, mosaic for a premutation. The mother carries a premutation allele with a $(CGG)_n$ repeat size of 101 and was identified as a carrier after her first son was diagnosed with fragile X syndrome (630 repeats). Analysis of direct CVS in the current pregnancy identified a male fetus. Molecular analysis revealed a fetus mosaic for FMR-1 alleles with $(CGG)_n$ repeat sizes ranging from 130-170. After counselling, the couple chose to pursue amniocentesis to confirm that the expansion in the extraembryonic tissue accurately reflected the somatic expansion size in the fetus and to assess FMR-1 methylation status. Amniocentesis revealed a male with an unmethylated premutation and a (CGG)n repeat size of 170 repeats. Based on these findings we predict that the fetus is unlikely to be affected with fragile X syndrome. At birth, cord blood will be analyzed to confirm the above findings. The majority of published recommendations regarding the identification of a male carrier of fragile X syndrome on CVS suggest confirmation of this result through amniocentesis or cordocentesis. Additional cases describing the accurate diagnosis of male carriers of fragile X syndrome by CVS need to be reported to facilitate the development of evidence-based protocols regarding the management of these cases. This could potentially alleviate the need for a second invasive prenatal procedure.

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Interactive web-based genetic screening questionnaires in a primary care and obstetrics practice: A pilot study. J.A. Neidich[1], C. Taswell[1] and K. Daniels[2]. [1]Global TeleGenetics, Inc., Palo Alto, CA, and [2]Stanford Health Services and Lucile Packard Children's Hospital, Stanford, CA.

As research on the genetic basis of disease builds our medical knowledge base, new developments in clinical genetics should also reach the patients whose health may benefit from such knowledge. Although primary care physicians and other healthcare providers usually act as the gateway to genetic consultation or testing, many providers have minimal formal training in genetics or otherwise may not be up-to-date in the field. Thus, they may not know when to refer a patient for genetic services.

To assist physicians and other healthcare providers in assessing whether a patient would benefit from genetic consultation, we have developed a series of web-based genetic screening questionnaires for use in primary care settings and obstetrics practices. The questionnaires provide a simple interactive format easily answered by the patients. The automated questionnaires then return an email message to the referring physician or healthcare provider and to the patient with a recommendation about the appropriateness of genetic consultation.

To test the acceptance by patients and providers of web-based genetic screening, we conducted a pilot study in a primary care setting linked to a major medical center. The patients who use this clinic represent all socioeconomic levels and racial and ethnic groups. The pilot study included twenty-five obstetric patients and twenty-five patients seen for other reasons. After the patients submitted their genetic screening questionnaires, they also completed separate questionnaires evaluating their experience. All medical personnel completed analagous questionnaires evaluating their experience with the overall process. The results of the study will be discussed.