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BRCA genetic testing: where are physicians in the decision process? C.M. Velicer[1,2], S.Taplin[1,3]. [1]Group Health Cooperative, Seattle, WA, [2]Univ. of WA, Dept. of Epidemiology, Seattle, [3]Univ. of WA, Dept. of Family Medicine, Seattle, WA.

Testing for mutations in hereditary breast/ovarian cancer genes (BRCA1/BRCA2) is appropriate for a limited group of high-risk individuals, such as some breast cancer survivors. It is not known if survivors obtain information regarding genetic testing or whether physicians play a role in the testing decision process. Our objectives were to determine if high-risk survivors speak with physicians and relatives about BRCA genetics, and to determine their knowledge, testing intentions, and preferred information sources. We conducted a population-based cross sectional study in 1/99 at Group Health Cooperative. All 276 female current enrollees initially diagnosed with breast cancer 5-10 years prior to 6/30/98 and 40-49 years old at diagnosis were mailed a survey. Of 217 respondents, 8% spoke with physicians and 53% spoke with relatives. On average, women correctly responded to 2 of 7 true/false questions. Respondents who spoke with physicians had higher knowledge scores than those who did not ($p<.001$). Respondents preferred written materials (80.6%) and discussions with physicians (79.8%) for BRCA information. In summary, survivors have limited BRCA knowledge, though they are discussing genetics with relatives. Physicians currently give little input regarding BRCA testing, but survivors would consider their involvement helpful.

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Evaluating data from newborn screening programs - Georgia, 1998. S.S. Wang[1], P.M. Fernhoff[2], K. Grinzaid[2], M. Ramachandran[3], E.A. Franko[3], M. Henson[4], J. Buehler[5], M.J. Khoury[1]. [1]Office of Genetics and Disease Prevention, CDC, Atlanta, GA, [2]Emory Univ. School of Medicine, Atlanta, GA, [3]Georgia Public Health Laboratory, Atlanta, [4]Georgia Genetics Program, Atlanta, [5]Div. of Public Health, GA.

Newborn screening (NBS) programs in the U.S. annually screen four million infants for metabolic and other disorders to prevent mental retardation, disability, and death. Assuring the effectiveness of NBS programs requires the collection of appropriate data for program evaluation, including short-term outcome measures (screening coverage, specimen quality, timeliness of diagnosis/treatment) and long-term outcome measures (morbidity, premature mortality, disability). We evaluated the data systems of the GA NBS program (GA NBS laboratory database and Emory University Medical Genetics database) and assessed our ability to measure these short- and potential long-term outcomes.

From January 1, 1998 to December 31, 1998, the NBS laboratory received 199,387 specimens. Of these specimens, 135,163 (67.8%) were collected in a satisfactory manner and received by the laboratory in the appropriate time frame (less than one week of age). Another 20,839 specimens (10.4%) were collected in a satisfactory manner, but obtained after one week of age. 20,691 (10.4%) specimens were collected from low birth weight newborns and 20,687 (10.4%) specimens were classified as unsatisfactory. Of all specimens, there were 4,557 initially abnormal screening results. Final normal results were ascertained for 4,094 of these results. Clinically significant disorders were diagnosed in 93 infants; these infants received their first abnormal test results within one week of age and initial treatments between one week to two months of age.

Data unavailable from the current system include the number of children the 199,387 specimens represent. Furthermore, the current system does not allow for the assessment of long-term follow-up outcomes on the 93 children diagnosed with clinically significant disorders. These results demonstrate that while some short-term outcomes are measured, screening coverage and effectiveness of the NBS program in preventing disabilities and/or premature death cannot be adequately determined. As with other state NBS programs, there is an urgent need for the collection and linkage of various sources of information in order to conduct program evaluations.

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Visual presentations at 1998 ACMG meeting suggests insensitivity of geneticists to a common genetic disorder. M.S. Williams[1], J.L. Williams2. 1Gundersen Lutheran Medical Center, 2La Crosse Regional Genetics Services Project, La Crosse, WI.

Red-green colorblindness (colorblindness, partial, deutran series, OMIM #303800) is a common X-linked recessive disorder affecting approximately 8% of the male population. Individuals with this disorder have difficulty perceiving the difference between the primary colors red and green and also experience difficulty perceiving these colors against certain backgrounds. Presentations at medical meetings are very visually oriented. The new computer slide programs allow for very complex and colorful transparencies to be easily created. Comments to the authors from acquaintances with red-green colorblindness regarding their difficulty reading slides that use the colors red and green led to the current study. Thirty-six presentations at the 1998 Annual Clinical Genetics Meeting of the ACMG were attended by one or both of the authors. For each session data was obtained on three parameters: Were slides used, were the colors red and/or green used in text or figures of the slides, how many slides used red and/or green to highlight information critical to the understanding of a slide. Only primary red or green were scored as a positive. Of the 36 presentations, 34 utilized slides. Of these 34 presentations, 19 (56%) used red and/or green. In all cases (19/19 100%) red and/or green was used to highlight information critical to the understanding of at least one slide in these presentations. The average number of critical slides containing red or green highlighted information was 3.1 per presentation. Geneticists should be aware that use of red or green in presentations impairs information transfer to a portion of their audience.