INVITED COMMENT

AMERICAN JOURNAL OF medical genetics PART A

Call for Change in Prenatal Counseling for Down Syndrome

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Received 11 September 2011; Accepted 23 November 2011

The *American Journal of Medical Genetics Part A* is to be congratulated for taking a leadership role by publishing a number of papers challenging the status quo of prenatal counseling for Down syndrome and of care for children and adults with Down syndrome. Parents want to know about the future abilities and potential of their fetus with Down syndrome, not simply negative medical information that may be outdated. Those providing counseling and those providing medical care could benefit from contact with individuals with Down syndrome outside the medical context. It is imperative that each person with Down syndrome be viewed as a unique individual with particular talents. Medical care providers should work with parents to help the child or adult with Down syndrome reach his/her goals. © 2012 Wiley Periodicals, Inc.

Key words: Down syndrome; prenatal testing; genetic counseling

How to Cite this Article:
McCabe LL, McCabe ERB. 2012. Call for change in prenatal counseling for Down syndrome.
Am J Med Genet Part A 158A:482–484.

Pregnant women and their partners who are deciding whether or not to pursue any prenatal testing or additional prenatal testing for Down syndrome deserve accurate information regarding the quality of life for an individual with Down syndrome. This information should be as current as possible and free from any bias of the individual or the agency presenting the information. The *American Journal of Medical Genetics Part A* has recently provided a great service to the genetics community by publishing a number of articles addressing this issue.

Surveys of parents of children with Down syndrome and of genetic counselors identified 34 essential pieces of information for the initial discussion of Down syndrome [Sheets et al., 2011a]. Genetic counselors were more likely to emphasize clinical information and negative aspects of the diagnosis, while parents valued information regarding the abilities and potential of individuals with Down syndrome. Fifty-six and eight-tenths percent of the parents felt that they were not adequately informed regarding Down syndrome at the time of diagnosis while 50.5% of the parents were somewhat or extremely dissatisfied with the experience of receiving their child's diagnosis. Similarly, Van Riper and Choi [2011] reported that the current level of parental satisfaction with the informing process, 52% for Mothers and 50% for Fathers, has not changed in the past 20 years.

Those providing genetic counseling need to be aware of the experience of individuals with Down syndrome, their parents and their siblings [Skotko et al., 2011a,b,c]. Individuals with Down syndrome are happy with their lives (99%), like who they are (97%), like how they look (96%), love their families (99%), like their siblings (97%), and feel that they make friends easily (86%) [Skotko et al., 2011c]. Parents with a son or daughter with Down syndrome report that they love their child (99%), are proud of them (97%), and feel their outlook on life is more positive because of them (79%) [Skotko et al., 2011b]. Parents also report that their typical children have good relationships with their child with Down syndrome (95%) [Skotko et al., 2011b]. Only 4% of parents of a child with Down syndrome regret having them and only 5% are embarrassed by their child with Down syndrome. Siblings of an individual with Down syndrome have affection for their sibling with Down syndrome (96%), are proud of their sibling (94%), feel they are a better person because of their sibling (88%), and plan to remain involved in their sibling's life into adulthood (90%) [Skotko et al., 2011a]. Less than 10% of siblings feel embarrassed by their sibling with Down syndrome and only 5% would trade their sibling with Down syndrome for one without Down syndrome. Therefore, individuals with Down syndrome value themselves, and their

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Grant sponsor: Anna and John J. Sie Foundation; Grant sponsor: Global Down Syndrome Foundation.

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DOI 10.1002/ajmg.a.35197

parents and siblings feel that these individuals are important members of their family.

Those providing genetic counseling should increase their contact with individuals in the disabilities community outside the medical context to understand and to be able to describe knowledgably what the lives of people with disabilities are really like. The relationship between genetic counselors and the disability community has been questioned by a group of genetic counselors [Madeo et al., 2011]. These authors recommend that the National Society of Genetic Counselors (NSGC) facilitate interactions between genetic counselors and members of the disabilities communities at events such as the NSGC Annual Education Conference, through NSGC funding for research involving individuals with disabilities, and by including disability rights in the NSGC strategic plan. They also recommend that the *Journal of Genetic Counseling* have a special issue on disability rights. They suggest that the American Board of Genetic Counseling revise their competencies to include issues relevant to the disability communities. They recommend that training program accreditation consider student study of disability rights and participation in the disability community beyond the clinical setting. The authors indicate that consumers of genetic services should be invited to participate on accreditation site visit committees.

Genetic counseling could be improved by following the new NSGC practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome [Sheets et al., 2011b]. In addition to genetic counselors, physicians could benefit from training regarding Down syndrome according to Pace et al. [2011], whether they are providing counseling or medical care. In a survey of physicians, 18.4% agreed with the statement that children with Down syndrome should attend special schools and 29.8% were neutral, totaling nearly 50% who did not advocate for inclusion. 24.3% agreed with the statement that children with Down syndrome are distracting in a classroom with typically developing students and 36.7% responded neutrally, or >60% accepting the concept that these children distract the typical children in the classroom, a position not supported by data [Loreman, 2007; Jackson et al., 2008–2009; Ruijs et al., 2010]. 9.8% did not feel comfortable providing medical care to a person with Down syndrome and 14.3% were neutral for a total of >25%. 1.9% felt that adults with Down syndrome were unable to work and 9% were neutral or >10%, when there is considerable evidence that people with Down syndrome are part of the workforce [Alderson, 2001; Smith, 2001].

An illustration of the timeliness and the need for these publications is seen in the materials used by the California Prenatal Screening Program as discussed by McCabe and McCabe [2011]. The California Prenatal Screening Program's brochure for couples with a first trimester (CPSP: Results for Screening in the First Trimester, March 2009) or second trimester (CPSP: Results for Screening in the Second Trimester, April 2009) positive screen for Down syndrome erroneously state: "Infants with this birth defect are moderately retarded; a few are mildly or severely retarded." Another publication of the California Prenatal Screening Program, the clinical Provider Book, also describes "most individuals" with Down syndrome as "moderately retarded" (Appendix C; CPSP: Provider Book, March 2009). These materials for parents and medical professionals overestimate the percentage of individuals with Down syndrome and moderate intellectual disability, and underestimate the percentage of individuals with Down syndrome and mild intellectual disability. Carr [1995] reported that 34% of individuals with Down syndrome had IQs in the mild disability range. Jamie Edgin [Personal Communication] replicated these findings by showing 39.4% of individuals with Down syndrome had mild intellectual disability and 1% had borderline intellectual functioning. In addition, the materials use the terminology "retarded," which the intellectual and developmental disabilities advocacy community considers insensitive and objectionable (R-Word, http://www.r-word.org/). In fact, on October 5, 2010, President Obama signed Rosa's Law which amends language in federal health, education and labor laws to remove the words "mentally retarded" and to replace them with "intellectual disability" (Remarks by the President at the Signing of the 21st Century Communications and Video Accessibility Act of 2010).

While we are discussing these brochures, they do not present all the options available. In the first trimester brochure, for the response to the question, "What can you do next?," the only options listed are: "1. Another blood screening test between 15 and 20 weeks to get a revised risk;" or "2. Follow-up testing to know for sure if the fetus has Down syndrome." Likewise the second trimester brochure's answer is "Follow-up testing to know for sure if the fetus has Down syndrome." There is not an option indicating no further testing. There is clearly an implication that further testing is necessary. Mothers and their partners have the right to decline testing.

In their response to our commentary, Lorey and Flessel [2011], representing the California Prenatal Diagnosis Program, did not take responsibility for the incorrect information in their materials. To assist the genetic counselor or perinatologist at the California Prenatal Diagnostic Centers or the woman's physician in presenting information about Down syndrome, we would recommend to Lorey and Flessel that they share the recent publications cited in this paper [Madeo et al., 2011; Sheets et al., 2011a,b; Skotko et al., 2011a,b,c] with the individuals responsible for prenatal counseling and that these papers be used to rewrite the entries regarding Down syndrome in their patient booklets and provider book. As one of the states with the highest birth rates, California could play an important part in changing prenatal counseling for Down syndrome.

By interacting with individuals with Down syndrome and their families, those who provide prenatal diagnosis counseling could learn that individuals with Down syndrome consider themselves to be actors, artists, athletes, daughters, friends, siblings, sons, students, volunteers, and workers. They might also discover that grandparents, parents, and siblings cherish their special relationships with an individual who provides them with an opportunity they would not have missed for anything.

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