

The California Prenatal Screening Program: “Options and choices” not “coercion and eugenics”

Monica C. Flessel, PhD¹, and Fred W. Lorey, PhD²

Abstract: The California Prenatal Screening Program is designed to make prenatal screening available to the state’s large and diverse population. The Program provides information to women which will allow them to make informed choices regarding prenatal screening and prenatal diagnosis. Since the Program’s inception in 1986, women in California have had the option to participate in prenatal screening or to decline prenatal screening. The California Program offers prenatal diagnostic services to women whose screening tests indicate an increased risk for birth defects, including Down syndrome. Women can decline any or all of these follow-up services. Genetic counseling, diagnostic services, and the presentation of diagnostic results are performed by medical professionals (not State staff) who follow established guidelines for nondirective counseling. Program data clearly demonstrate that women in California have a wide range of options and make a wide range of choices regarding prenatal screening and prenatal diagnosis. California’s comprehensive Prenatal Screening Program promotes optimal care for all women within all options and choices. The important and necessary communication among organizations and stakeholders involved in prenatal screening and diagnosis, and in related care for pregnant women and for people with Down syndrome, is not served by misrepresentation and inflammatory rhetoric. *Genet Med* 2011;13(8):711–713.

Key Words: California Prenatal Screening Program, Down syndrome, trisomy 21, prenatal diagnosis, patient choice

THE CALIFORNIA PRENATAL SCREENING PROGRAM

California and Iowa are the only two states in the United States that perform prenatal screening as part of a statewide public health program. The California Program is designed to make prenatal screening available to our large and diverse population. The Program strives to provide information to women which will allow them to make informed choices regarding prenatal screening and prenatal diagnosis. Since the Program’s inception in 1986, women in California have had the option to participate in prenatal screening or to decline prenatal screening. Since 2009, the Program has included options for first and/or second trimester screening. Currently, about 400,000 women per year participate in the Program, corresponding to approximately 75% of pregnant women in California (unpublished Program data).

The California Program is unique in offering prenatal diagnostic services to women whose screening tests indicate an

increased risk for birth defects, including Down syndrome. Women can decline any or all of the follow-up services offered through the Program at Prenatal Diagnostic Centers (PDCs). Those services begin with counseling by a professional genetic counselor, while subsequent services include ultrasound, chorionic villus sampling, or amniocentesis. Table 1 shows data from the California Prenatal Screening Program for women with screen positive results for Down syndrome. Approximately 77% of women at increased risk for Down syndrome accept a referral to a PDC. Essentially, all of these women receive genetic counseling and nearly all (99%) of the women seen in the second trimester accept the offer of ultrasound. Only about 43% of women at the PDCs choose diagnostic testing through chorionic villus sampling or amniocentesis. The California Program covers PDC services for patients up through the completion of diagnostic testing. The services are provided by PDC staff, such as genetic counselors and perinatologists, not State staff. For a pregnancy with a diagnosis of a birth defect or pregnancy complication, the patient’s decisions about pregnancy management and pregnancy options are made in consultation with medical professionals, outside of the Program.

Clearly, there are many advantages to prenatal diagnosis of Down syndrome for women (and families) who continue the pregnancy. Families have the opportunity to learn more about Down syndrome and to connect with support groups. They have the ability to plan ahead for the birth and to arrange for delivery at a medical center with specialists available to assess the baby’s medical condition. Table 2 shows recent California data confirming that a higher percentage of prenatally diagnosed cases compared with undiagnosed cases delivered at regional medical centers (27.4% vs. 19.2%) and a lower percentage of diagnosed cases compared with undiagnosed cases delivered at community hospitals lacking a neonatal intensive care unit (23.8% vs. 30.2%). Providing opportunities to improve the birth experience and the birth outcome for families with a Down syndrome baby is an important aspect of the California Program.

“CHOICES AND OPTIONS” NOT “COERCION AND EUGENICS”

We are concerned that the Commentary by McCabe and McCabe¹ misrepresents the mission and philosophy of the California Program. Thus, we feel it is important to respond to the serious charges of coercion and eugenics leveled against the Program, clarify the status of prenatal screening and diagnosis of Down syndrome in California, and further describe the Program’s many options and choices available to patients.

In the Commentary by McCabe and McCabe,¹ their discussion of the California Program begins with the statement “Pressure is exerted by governmental entities to terminate pregnancies when prenatal testing indicates a fetus with Down syndrome.” This is not true. Not a single example of such purported pressure from the California Program to terminate pregnancies is described. What does follow the Commentary

From the ¹Prenatal Screening Branch, ²Genetic Disease Screening Program, California Department of Public Health, Richmond, California.

Monica C. Flessel, PhD, Genetic Disease Screening Program, California Department of Public Health, 850 Marina Bay Parkway, F-175, Richmond, CA 94804. E-mail: monica.flessel@cdph.ca.gov.

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Table 1 Patient choices of referral and diagnosis from April, 2009 to March, 2010

Screen positive for Down syndrome	20,927
Accepted referral to PDC	16,088
Screen positive accepting PDC referral (%)	77%
Chose diagnostic procedure at PDC	6961
Chose diagnostic procedure at PDC (%)	43%

Table 2 Impact of prenatal diagnosis on the choice of birth hospital, 2005–2007

	Hospitals without NICU n (%)	Community referral, hospital n (%)	Regional referral hospital n (%)	Total
Down syndrome births				
No PDC referral	129 (30.2)	216 (50.6)	82 (19.2)	427
PDC referral, no diagnosis procedure	123 (28.9)	203 (47.7)	100 (23.5)	426
PDC referral, diagnosis performed	40 (23.8)	82 (48.8)	46 (27.4)	168

Cox-Mantel-Haenzel χ^2 for trend $P < 0.05$.
NICU, neonatal intensive care unit.

statement are two quotations from Program publications, taken out of context and misrepresented as to their actual intent.

The first quotation of “missed opportunities”² is presented by McCabe and McCabe as referring to diagnosed cases of Down syndrome where the pregnancy is continued. In fact, the paragraph from which the quote is taken describes the need to reduce the number of affected pregnancies that are not identified through prenatal screening by making improvements to the screening process and assays. McCabe and McCabe seem to take issue with the paper’s topic of cost effectiveness,² noting “The California program’s position is most definitely based on economic considerations—in terms of cost and effectiveness.” Certainly, cost effectiveness is an important component of healthcare and of public health programs. It is, however, only one of 10 criteria adopted for screening programs by the World Health Organization.³ The California Prenatal Screening Program is responsive to all 10 of the World Health Organization criteria and to all of the recently updated criteria for screening programs.⁴ The updated criteria include mandates for equity and access for the entire target population; informed choice; evidence-based screening; and quality assurance. Indeed, we consider all of these criteria to be hallmarks of our statewide Program.

The second quotation used in the Commentary is taken from the Program’s booklet for women who have a prenatal screening result indicating increased risk for Down syndrome.⁵ The Commentary fails to explain that the booklet is not a stand-alone document. The booklet is used by physicians and genetic counselors as a helpful handout for patients. The quotation in the Commentary comes from the page addressing the question: “What if Down syndrome is found?” The section quoted in the Commentary is as follows: “Infants with this birth defect are moderately retarded, a few are mildly retarded or severely retarded. Heart defects are common. These heart defects can

usually be treated with surgery and medication. Other serious health problems often exist with Down syndrome. Medical treatment can help some of these problems.”⁵ The Commentary authors express their concern that “the inaccurate and overly negative information provided by California may be intended to coerce a woman into a decision to terminate her pregnancy if the fetus is diagnosed with Down syndrome.”¹

To alleviate their concerns, the authors need only to have consulted the booklet text before and after the section they quoted. The sentence before their quotation is the first sentence that follows the question: “What if Down syndrome is found?” That sentence reads: “A doctor or genetic counselor would give you information about Down syndrome.”⁵ This reflects the fact that the diagnosis of Down syndrome is usually presented by a genetic counselor or perinatologist at the PDC, or else the patient’s doctor. The diagnosis and counseling are presented by medical professionals (not State staff) who follow established guidelines for nondirective counseling.

The patient booklet sentences that follow the Commentary quote read: “Special programs are available throughout California to help children and adults with Down syndrome to achieve their full potential. Options for continuing or ending the pregnancy will be discussed during counseling. The decision is entirely up to you.”⁵ We agree with the Commentary authors that patients receiving a prenatal diagnosis of Down syndrome should be given “the most neutral, nondirective, prenatal genetic counseling”¹ and we strive to be informative, nondirective, and neutral in our patient education materials, as well.

The two following quotations from the Commentary are presented as “factual” statements, with reference to the same citation. The first statement is “The California program’s rhetoric and brochure... threaten personal autonomy and reproductive freedom.” The second statement is “The rhetoric from the California prenatal testing program... is considered to facilitate eugenics.”¹ The citation for these declarative statements turns out to be a previous opinion piece by the same authors.⁶ In stark contrast, we believe Program data clearly demonstrate that women in California have a wide range of options and make a wide range of choices regarding prenatal screening and prenatal diagnosis. We would argue that our comprehensive prenatal screening program is designed to prevent an approach based on coercion and eugenics and promotes optimal care for all women within all options and choices. The web site for the California Prenatal Screening Program (www.cdph.ca.gov/programs/pns) contains all of our clinician and patient education materials and other Program information. The Program administrative staff welcomes review and comments from any interested members of the community.

CONTINUING NEED FOR “CONSENSUS CONVERSATION”

The final citation in the Commentary references the consensus document from a meeting designed to bring together a wide range of organizations involved in the prenatal screening and diagnosis of Down syndrome.⁷ Unfortunately, McCabe and McCabe seem to have based their Commentary on many of the misperceptions about Down syndrome that the “Consensus Conversation” was attempting to dispel. There is clearly a need for continued and open discussion among all organizations and stakeholders to address the complicated social and medical issues involved in prenatal screening and diagnosis and in related care for pregnant women and for people with Down

syndrome. However, misrepresentation and inflammatory rhetoric have no place in such discourse. An excellent example has been set for outreach and communication⁷ that we should all strive to follow.

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