State of California—Health and Human Services Agency
California Department of Public Health

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GENETIC DISEASE SCREENING PROGRAM

PRENATAL DIAGNOSIS CENTER

FINDING OF EMERGENCY

December 3, 2019

Health and Safety Code (HSC) Section 124977(d) authorizes the California Department of Public Health (Department) to adopt emergency regulations. The adoption of these regulations is deemed an emergency. These regulations are necessary for the immediate preservation of the health, safety, and general welfare of Californians.

HSC Section 124977(d)(1) states: "For the purposes of the Administrative Procedure Act, the adoption of regulations shall be deemed an emergency and necessary for the immediate preservation of the public peace, health and safety, or general welfare. Notwithstanding Chapter 3.5 (commencing with Section 11340) of Part 1 of Division 3 of Title 2 of the Government Code, these emergency regulations shall not be subject to the review and approval of the Office of Administrative Law. Notwithstanding Section 11346.1 and Section 11349.6 of the Government Code, the department shall submit these regulations directly to the Secretary of State for filing. The regulations shall become effective immediately upon filing by the Secretary of State. Regulations shall be subject to public hearing within 120 days of filing with the Secretary of State and shall comply with Sections 11346.8 and 11346.9 of the Government Code or shall be repealed."

HSC Section 124977(d)(2) states: "The Office of Administrative Law shall provide for the printing and publication of these regulations in the California Code of Regulations. Notwithstanding Chapter 3.5 (commencing with Section 11340) of Part 1 of Division 3 of Title 2 of the Government Code, the regulations adopted pursuant to this chapter shall not be repealed by the Office of Administrative Law and shall remain in effect until revised or repealed by the department."

Senate Bill (SB) 1555 (Speier, Chapter 484, Statutes of 2006) also added what is now HSC Section 125055(g) allowing the Department to adopt emergency regulations to implement and make specific the amendments made by SB 1555, expanding the California Prenatal Screening Program to include "all tests that meet or exceed the current standard of care as recommended by nationally recognized medical, or genetic organizations, including, but not limited to inhibin." HSC Section 125055(g) states "the regulations be deemed an emergency and necessary for the immediate preservation of the public peace, health and safety, or general welfare."

SUMMARY OF PROPOSAL

The purpose of these regulations is to implement, interpret, and make specific Senate Bill (SB) 1555 (Speier, Chapter 484, Statutes of 2006) and update obsolete terms and program terminology. SB 1555 added Health and Safety Code (HSC) section 125055(g)(1) to require that prenatal screening include "all tests that meet or exceed the current standard of care as recommended by nationally recognized medical or genetic organizations, including, but not limited to, inhibin." The addition of inhibin (dimeric inhibin A, a protein produced by the ovaries and fetal placenta) as a fourth analyte tested in maternal serum in the second trimester makes the screening more accurate in detecting pregnancies at risk of Down syndrome.

In accord with this legislative mandate, the California Department of Public Health (the Department) has also responded to recommendations that first trimester serum screening be offered to pregnant individuals as the standard of care; that cell-free DNA (cfDNA) screening be available to high-risk individuals as a follow-up service; and that prenatal microarray testing be available to individuals with findings of major structural anomalies on ultrasound. First trimester serum screening now involves the testing of a fifth analyte, pregnancy-associated plasma protein A, associated with fetal chromosomal anomalies including aneuploidy (an abnormal number of chromosomes). This regulatory action was required to reflect this program expansion and the current standards and procedures that contribute to the health and safety of individuals and children in California.

DOCUMENTS RELIED UPON

The following documents were relied upon in developing these regulations:

American Academy of Pediatrics and American College of Obstetricians and Gynecologists, 2007. *Guidelines for Perinatal Care (6th ed.)*, Elk Grove Village, American Academy of Pediatrics.*

American College of Obstetricians and Gynecologists, "Screening for Fetal Chromosomal Abnormalities," *ACOG Practice Bulletin – Clinical Management Guidelines for Obstetrician-Gynecologists*, Number 77, January 2007.*

American College of Obstetricians and Gynecologists Committee on Genetics and The Society for Maternal-Fetal Medicine Publications Committee, "Noninvasive Prenatal Testing for Fetal Aneuploidy," *Committee Opinion,* Number 545, December 2012.

Available at:

http://www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-on-Genetics/Noninvasive-Prenatal-Testing-for-Fetal-Aneuploidy

American College of Obstetricians and Gynecologists Committee on Genetics, "The Use of Chromosomal Microarray Analysis in Prenatal Diagnosis," *Committee Opinion*, Number 581, December 2013.

Available at:

http://www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-on-Genetics/The-Use-of-Chromosomal-Microarray-Analysis-in-Prenatal-Diagnosis

American College of Obstetricians and Gynecologists Committee on Genetics and The Society for Maternal-Fetal Medicine, "Cell-free DNA Screening for Fetal Aneuploidy," Committee Opinion, Number 640, September 2015.

Available at:

http://www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-on-Genetics/Cell-free-DNA-Screening-for-Fetal-Aneuploidy

Driscoll, D.A., and Gross, S.J., "First trimester diagnosis and screening for fetal aneuploidy," American College of Medical Genetics Practice Guidelines, *Genetics IN Medicine*, January 2008, Vol. 10, No. 1: 73-75.

Available at:

http://www.acmg.net/StaticContent/SGs/First_trimester_diagnosis_and_screening_for_fetal.10.pdf

Gregg, A.R., Gross, S.J., Best, R.G., Monaghan, K.G., Bajaj, K., Skotko, B.G., Thompson, B.H., and Watson, M.S.; The Noninvasive Prenatal Screening Work Group of the American College of Medical Genetics and Genomics, "ACMG statement on noninvasive prenatal screening for fetal aneuploidy," *Genetics in Medicine*, May 2013;15(5): 395-8.

Available at:

https://www.acmg.net/docs/nips-GiM_galley_text_130301.pdf

*Viewing and Copies:

Upon request, the Department's Office of Regulations will have these documents available for viewing during the public comment period(s).

Prenatal Diagnosis Center Standards and Definitions 2018

American College of Medical Genetics and Genomics, Standards and Guidelines for Clinical Genetics Laboratories, 2018 Edition, Revised January 2018

AUTHORITY

The Department is proposing to adopt, amend, or repeal, as applicable, the proposed regulations under the authority provided in HSC Sections 124977, 124980, 124996, 125000, 125055, 125070 and 131200.

This proposal implements, interprets, and makes specific HSC Sections 1367.54, 124975, 124977, 124980, 124990, 124996, 125000, 125001, 125050, 125055, 125060, 125065, 125070, 125075, 125085, and 131052, and Insurance Code Section 10123.184.

INFORMATIVE DIGEST

Health and Safety Code (HSC) Section 125050 requires the California Department of Public Health (Department) to administer a statewide program for prenatal testing for genetic disorders and birth defects, including but not limited to ultrasound, amniocentesis, chorionic villus sampling, and blood testing. Testing requirements for prenatal birth defects are established in Title 17, California Code of Regulations (17 CCR), Section 6521, et seq.

POLICY STATEMENT OVERVIEW

Problem Statement: Senate Bill (SB) 1555 (Speier, Chapter 484, Statutes of 2006) added HSC Section 125055(g)(1) which requires the Department to expand prenatal screening and testing to meet the current standard of care. Regulatory action was required to implement, interpret, and make specific this provision relating to prenatal screening and testing in California by specifying this standard of care and the options offered to women who participate in the California Prenatal Screening (PNS) Program. Additional analytes in maternal serum are now tested in the first and/or second trimester as part of the California PNS Program (originally known as the California Alpha-fetoprotein [AFP] Screening Program, and subsequently as the Expanded AFP Screening Program). The Department has also responded to recommendations that cell-free DNA (cfDNA) screening is available to high-risk women as a follow-up service, and that prenatal microarray testing is available to women with findings of major structural anomalies on ultrasound through the California PNS Program.

With the program expansion, the regulation text contained obsolete terms, program references, standards, and clinical methods. Definitions were no longer accurate and referred to acronyms no longer associated with the new program name. The regulation text also contained inaccuracies and inconsistent uses of terminology.

Objectives (Goals): Broad objectives of this regulatory action are to:

Implement, interpret, and make specific HSC Section 125055(g)(1).

Specify the current standard of care for prenatal screening, as recommended by the American College of Obstetricians and Gynecologists (ACOG) and American College of Medical Genetics (ACMG) and offered by the California PNS Program. This includes additional analytes tested (dimeric inhibin A and pregnancy-associated plasma protein-A) and the screening options and follow-up services available for women participating in the California PNS Program.

Update the program name by replacing references to the “Expanded AFP Program” with the “California Prenatal Screening Program.”

Update the period for drawing blood samples and performing serum screenings to include the first trimester.

Update the clinical process used to assess fetal age.

Repeal or update obsolete and inaccurate definitions, terminology, and methods, including repeat testing for screen positive results.

Ensure accurate and consistent use of terminology and definitions.

Update requirements for State-approved Prenatal Diagnosis Centers (PDCs), personnel and laboratories providing authorized follow-up services to women participating in the California PNS Program.

Benefits:

Anticipated benefits, including nonmonetary benefits, from this regulatory action are to:

Protect the health, safety, and welfare of women and children by specifying the standard of care for prenatal screening and testing offered through the California PNS Program.

Reduce confusion for the regulated community, healthcare providers, the Department's vendors, third party payers, and the general public by removing or updating obsolete terminology, methods, definitions, and standards.

Provide clarity in the use of terminology, definitions, and procedures authorized by the Department.

OTHER STATUTORY REQUIREMENTS

None.

DETERMINATION OF LOCAL MANDATE

The Department has determined that the emergency rulemaking does not impose a mandate on local agencies or school districts, nor are there any costs for which reimbursement is required by Part 7 (commencing with Section 17500) of Division 4 of the Government Code.

FISCAL IMPACT ESTIMATES

Cost to any local agencies or school districts that must be reimbursed pursuant to Section 17561 of Government Code:

None.

The cost or savings to any state agency:

The Department estimates there will be no impact on the Genetic Disease Testing Fund. SB 1555 increased the California PNS Program participation fee by $40 for the mandated program expansion, and by an additional $10 to support the pregnancy blood sample storage, testing, and research activities of the Birth Defects Monitoring Program. This $50 fee increase was reflected in an amendment to 17 CCR, Section 6540 in 2007, pursuant to Title 1, California Code of Regulations, Section 100. A subsequent $7 fee increase was enacted through regulatory action in 2011 to maintain program solvency,

followed by a $45 fee increase from July 1, 2014, to correct for overstatements of estimated caseloads and revenue reductions from changes to the Medi-Cal reimbursement rate that had led to a cumulative deficit in GDTF. On July 1, 2016 the fee increased by $14.60 to a total of $221.60 to cover additional administrative program costs. These fee increases were necessary to maintain the self-sufficiency of the California PNS Program.

Other nondiscretionary costs or savings imposed on local agencies:

None.

Impact on any cost or savings in federal funding of the program:

None.