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Laboratories

February 25, 2014

To: Ms. Kirstin Michel
Department of Health Care Policy and Financing
Colorado Medicaid
kirstin.michel@state.co.us

RE: Request for HerediT™ Cystic Fibrosis Carrier Screen Coverage by Colorado Medicaid

Dear Ms. Michel,

Sequenom Laboratories respectfully requests coverage and appropriate payment for Cystic Fibrosis carrier screening performed by our certified and accredited laboratory under the name HerediT™. A review of the information provided below and the referenced information which is attached may be helpful concerning your consideration.

**Cystic Fibrosis (CF) Carrier Screening**
Cystic fibrosis (CF) is the most common life-threatening autosomal recessive condition in North America. It is a progressive, multisystem disease primarily affecting the pulmonary, pancreatic and gastrointestinal systems. Average life expectancy is 37 years.[1] In accordance with the ACOG Committee Opinion No. 486 published in 2011, all women of reproductive age should be offered carrier screening for Cystic Fibrosis.[1]

**Clinical Utility: Provides meaningful information to clinicians for counseling patients**
The Heredi-T CF test from Sequenom is a laboratory-developed test that analyzes 136 mutations and 5 variants that have been proven to be clinically relevant. It is a simple blood or buccal (cheek) swab test and results are typically available within one week. Detection rates are increased over standard CF panels for individuals of all ethnic backgrounds. The test examines phenotypically relevant (disease-causing) mutations that were selected from the Johns Hopkins CFTR2 database (http://www.cftr2.org). The CF and CF associated disease-causing mutations were identified by collecting data from North American and European patient registries. As a result, clinicians utilizing the Heredi-T CF test are provided with clear results when counseling their patients about the risk to have an affected child. This allows patients to make appropriate reproductive decisions about prenatal testing with clinically reliable information. Other CF panels often include mutations of uncertain significance, making it difficult for patients and clinicians to understand the true risk of having an affected child. Carrier screening for cystic fibrosis is ideally performed in the preconception period to allow couples to consider a complete range of pregnancy options, including the decision to conceive, preimplantation genetic diagnosis, prenatal testing or donor gametes.[1]

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**Expert Support for Implementation**
Most state funded programs and private health insurers support coverage for CF carrier screening for all patients who are pregnant or considering pregnancy in accordance with the 2011 ACOG guideline. Additionally, other professional organizations such as the National Society of Genetic Counselors and the American College of Medical Genetics and Genomics also support carrier screening for women of reproductive age.[2-3]

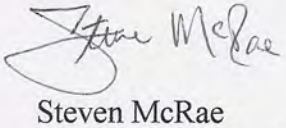
**Evidence**
Prenatal and Preconception screening for CF has been a part of routine obstetric and gynecology care since 2001.[1] Recently, it was suggested that mutation panels for carrier screening should be revised to include the disease causing mutations well characterized in the Johns Hopkins CFTR2 database.[4] The direct benefit in utilizing a panel suggested by CFTR2 for couples undergoing carrier screening is an increased sensitivity for detecting couples at 25% risk for an affected child.[4] Sequenom's HerediT CF panel is the only commercially available panel based on the CFTR2 database of clinically relevant mutations.

**Company Background**
Sequenom Laboratories is a life sciences company providing innovative genetic analysis solutions to improve healthcare. Our molecular diagnostic reference laboratory performs molecular diagnostic testing services utilizing its laboratory developed tests (LDTs). Our laboratory sites are accredited by the College of American Pathology, or CAP, and compliant with the certification requirements for high complexity testing under the Clinical Laboratory Improvement Amendments, of 1988, as amended, or CLIA. Patient samples are collected by health care professionals and submitted to Sequenom for testing and test results are reported back to the ordering physician. Sequenom provides high complexity testing to Medicaid patients nationwide and participates in 33 other state Medicaid programs.

A coverage and reimbursement pathway for this test incorporating CPT code 81220 will give Medicaid beneficiaries access to medically necessary testing in accordance with medical guidelines. It will also allow Medicaid beneficiaries to receive CF test results with clear clinical utility and increased detection rates and avoid findings of unclear significance.

Thank you for your time and for your consideration of this matter. Should you have any questions, please do not hesitate to contact myself or our Chief Medical Officer, Allan T. Bombard, MD, FACOG, FACMG at (858) 202-9027.

Sincerely,



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References


1. Update on carrier screening for cystic fibrosis. ACOG Committee Opinion No. 486. American College of Obstetricians and Gynecologists. Obstet Gynecol. 2011;117:1028-1031
2. Langfelder-Schwind, E, et al. Molecular Testing for Cystic Fibrosis Carrier Status Practice Guidelines: Recommendations of the National Society of Genetic Counselors. *J of Genetic Counseling.* February 2014, Volume 23, Issue 1, pp 5-15.
3. Watson MS, et al. Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel. *Genet Med.* 2004;6(5):387-391.
4. Sosnay, PR, et al. Defining the disease liability of variants in the cystic fibrosis transmembrane conductance regulator gene. *Nature Genetics.* 2013; 45(10):1160-7.