ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

March 2, 2022

Maria Finnell, MD
Chief Medical Officer
Indiana Family and Social Services Administration
402 West Washington Street
Indianapolis, IN 46204

Re: Indiana Medicaid Genetic Testing Policy

Dear Dr. Finnell,

I write to you on behalf of the American College of Medical Genetics and Genomics, the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics and the only medical specialty society in the US that represents the full spectrum of medical genetics disciplines in a single organization. ACMG strives to improve health through the clinical and laboratory practice of medical genetics as well as through advocacy, education, and clinical research, and to guide the safe and effective integration of genetics and genomics into all of medicine and healthcare, resulting in improved personal and public health. In line with our mission, we are aware that the Indiana Family and Social Services Administration (FSSA) is currently reviewing their prenatal genetic testing policies and provide the following information for consideration.

We were pleased to see that Indiana Medicaid mentions ACMG as a resource for test-specific criteria in its Genetic Testing policy. However, it is our understanding that current Indiana Medicaid policies specific to carrier screening, noninvasive prenatal screening using cell-free DNA (NIPS), and chromosomal microarray (CMA) during pregnancy may not reflect current professional recommendations. Lack of clearly defined and up-to-date policies can result in challenges for providers to obtain coverage and create barriers in access to standard testing for Indiana Medicaid beneficiaries. Without coverage, patients may have to pay out of pocket or forgo testing altogether.

Carrier screening is used to identify individuals or couples that are at risk to have a child with an autosomal recessive or X-linked genetic disorder, and those screened may use the results in their reproductive decision-making or to improve outcomes for their children. Carrier screening has been a routine component of

7101 Wisconsin Avenue
Suite 1101, Bethesda, MD 20814
Telephone: 301-718-9603
Fax: 301-718-9604

www.acmg.net

obstetrical care since the early 2000s. In a recently published practice resource[1], ACMG outlined four tiers of carrier screening. Specifically, ACMG recommends that Tier 3 carrier screening, which includes screening for over 100 serious inherited conditions, be offered to all pregnant patients and those planning a pregnancy. As such, prior authorizations, documentation of medical necessity, and results from diagnostic tests are not needed prior to Tier 3 carrier screening. Additional screening is recommended for pregnancies that stem from a known or possible consanguineous relationship or when otherwise warranted by a family or personal medical history. The ACMG recommendations ensure equitable care for a diverse U.S. population.

NIPS is used to identify pregnancies at risk of being affected by certain genetic conditions. Current evidence strongly suggests that NIPS can replace other conventional screening methods for certain chromosome abnormalities for pregnant patients regardless of their age or other risk factors, including aneuploidies such as Patau syndrome (trisomy 13), Edwards syndrome (trisomy 18), and Down syndrome (trisomy 21). In a 2016 position statement[2], ACMG recommended that all pregnant women be informed that NIPS is the most sensitive screening option for traditionally screened aneuploidies and informing them of the availability of the expanded use for screening for sex chromosome aneuploidies. ACMG also recommends allowing patients to select diagnostic or screening approaches for the detection of fetal aneuploidy and/or genomic changes that are consistent with their personal goals and preferences. For patients choosing NIPS, pre- and post-test counseling should be accessible, and appropriate diagnostic testing should be offered following a positive NIPS result. Since aneuploidy screening is a routine component of pregnancy management, a prior authorization requirement for this test would impose a significant administrative burden on already busy obstetricians with limited time for each patient.

CMA is used to detect chromosomal copy number changes, including deletions or duplications that are too small to be detected by conventional cytogenetics (known as microdeletion/microduplication syndromes). Many of these syndromes result in severe phenotypes after birth but have no discriminatory prenatal signs. Furthermore, women of all ages have equal risk for affected pregnancies. When there is suspicion for a microdeletion in a fetus, either from a NIPS result, abnormal ultrasound, or other personal or family history, CMA is the appropriate diagnostic test to confirm the diagnosis. In our 2016 position statement[2], ACMG recommended offering patients the option of fetal diagnostic testing (e.g., chorionic villous sampling or amniocentesis) followed by CMA using fetal DNA to maximize the detection of fetal genetic diagnoses. In the setting of fetal anomalies and a negative NIPS screening test, CMA has the highest diagnostic yield. For patients who have received a NIPS result indicating high-risk for a microdeletion syndrome, CMA is the appropriate diagnostic test to confirm the diagnosis.

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[1] Gregg AR, Aarabi M, Klugman S, Leach NT, Bashford MT, Goldwaser T, Chen E, Sparks TN, Reddi HV, Rajkovic A, Dungan JS; ACMG Professional Practice and Guidelines Committee. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2021 Oct;23(10):1793-1806. doi: 10.1038/s41436-021-01203-z. Epub 2021 Jul 20. Erratum in: Genet Med. 2021 Aug 27;: PMID: 34285390; PMCID: PMC8488021.

[2] Gregg AR, Skotko BG, Benkendorf JL, Monaghan KG, Bajaj K, Best RG, Klugman S, Watson MS. Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics. *Genet Med*. 2016 Oct;18(10):1056-65. doi: 10.1038/gim.2016.97. Epub 2016 Jul 28. PMID: 27467454.

As Indiana Medicaid evaluates its current coverage policies, we urge you to consider ACMG's carrier and prenatal screening recommendations as well as recommendations for prenatal CMA testing. We encourage Indiana Medicaid to develop clear coverage policies that reflect current professional recommendations for carrier screening, NIPS, and CMA and ensure equitable access for Indiana Medicaid beneficiaries.

For questions or additional information, please contact Michelle McClure, PhD, ACMG Director of Public Policy at mmcclure@acmg.net.

Sincerely,

Marc Williams, MD, FACMG
President
American College of Medical Genetics and Genomics