December 16, 2019

Neil Romano
Chairman
The National Council on Disability
1331 F St., NW, Suite 850
Washington, DC 20004

**Re: Council Recommendations on Genetic Testing**

Dear Mr. Romano:

While we appreciate the National Council on Disability's recent attention and focus on the issue of genetic testing, including noninvasive prenatal screening, we write to you today regarding several unsubstantiated claims and far-reaching recommendations included in the Council's recent report, *Genetic Testing and the Rush to Perfection*. As such, the report fails to reflect the highly regulated nature of the industry and clinical laboratories' commitment to care for individuals both with and without disabilities.

The American Clinical Laboratory Association (ACLA) is a not-for-profit association representing the nation's leading clinical and anatomic pathology laboratories, including national, regional, specialty, hospital, ESRD, and nursing home laboratories. A number of ACLA's clinical laboratory members offer genetic testing, including prenatal testing, and employ highly trained genetic counselors as a benefit to patients and a resource for physicians.

As your report notes, over the past several decades, scientific, medical, and technological advancements have enhanced our ability to treat the most complex health conditions facing patients, and genetic testing continues to play a fundamental role in our progress towards more personalized medicine. Importantly, we strongly support federal protections for individuals with disabilities and additional funding for expanded genetic testing education and for counseling in public health programs. ACLA and our members have also long advocated for a new regulatory framework for laboratory developed tests and diagnostics to enable patients to continue to have access to innovative, accurate, and reliable tests for their health.

Our members are committed to enabling providers and patients to have access to meaningful clinical data to better inform treatment plans. Many of the report's conclusions and recommendations — while intended to protect individuals with disabilities — ignore critical safeguards and federal and state regulations that promote testing accuracy and reliability as well as well-established conflict of interest standards for counselors. As a result, many of the report's conclusions and recommendations, if adopted, could have unintended harmful effects on individuals with and without disabilities.

With that in mind, we offer insight on three key areas, beginning with the value of non-invasive prenatal screening (NIPS) for patients who chose to receive this test. Further, we also believe it's important to underscore the consequences of haphazard interventions by the Food and Drug Administration (FDA), which would undermine ongoing efforts to advance comprehensive diagnostic reform in Congress — a priority for our organization and a necessity for maintaining patient access and innovation in our field. Finally, in response to the Council's concerns over conflicts of interest, this letter provides context on the rigorous compliance programs required of and implemented by labs that employ genetic counselors, which foster adherence to federal and state fraud and abuse laws, in

addition to professional codes of ethics. These existing measures are designed to protect patients, boost professional integrity, and prevent the alleged conflict that the council suggests.

### *Value of Non-Invasive Prenatal Screening (NIPS)*

While the Council's report outlines concerns over the growing use of NIPS, including its effects on women facing decisions about a current pregnancy, the American College of Obstetricians and Gynecologists (ACOG) recommends all pregnant women be offered aneuploidy screening.[1] NIPS is one mechanism to screen for aneuploidy, and importantly, represents an advance in standard of care, as the most accurate method of aneuploidy screening, which reduces the risk for invasive testing and thereby improves safety for both the mother and fetus.

Over the past decade, prenatal care has been revolutionized with the introduction of low-cost, blood based screening methods that provide expecting mothers, from 9 weeks gestational age, with vital information about serious birth defects, some of which (i.e., Trisomy 13 and 18) are often fatal. As more studies and clinical experience continue to demonstrate the efficacy of NIPS, this advanced screening technology has been recommended by ACOG and the American College of Medical Genetics and Genomics (ACMG), and it is now covered for all pregnant women by a significant and growing number of health insurance plans.

Women who cannot access NIPS as a first line screen for chromosomal abnormalities often instead undergo "maternal serum screening" (MSS), a testing technology that has been surpassed in efficacy by NIPS. A 2015 study published in the *New England Journal of Medicine* found that, for the most common chromosomal anomaly, Trisomy 21 (i.e. Down syndrome), MSS misses about 21% of pregnancies in which the anomaly is present, and also wrongly tells approximately 5% of expecting mothers that they tested "high risk" for Trisomy 21, even though 97% of those positive results are wrong.[2] The clinical consequence of these false positive results is that women will be unnecessarily offered invasive testing — amniocentesis or chorionic villus sampling — to establish a diagnosis; these invasive tests carry a small risk for miscarriage. It is contrary to the best interests of patient care to recommend that they utilize an inferior test when a superior option — NIPS — is readily available; yet that is the outcome that the Council's recommendations would dictate.

There are approximately six million annual U.S. pregnancies.[2] Without NIPS, many pregnant women will continue to be advised to undergo amniocentesis, an invasive procedure that carries potential risks, and which in turn causes hundreds of unnecessary pregnancy losses per year.[3] Ultimately, increase in cost and the potential loss of wanted pregnancies would be the result of policies that do not provide access to NIPS as a first-line screen for any woman who chooses to learn more about her baby. In addition, these patients should have the option to obtain guidance from licensed, certified genetic counselors to help them fully understand their test results and the treatment options that may be available.

ACOG has been joined in supporting NIPS by ACMG, which states: "[n]ew evidence strongly suggests that NIPS can replace conventional screening for Patau, Edwards, and Down syndromes across the maternal age spectrum."[4]

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[1] *See* Gregg AR, Skotko BG, Benkendorf JL *et al.* Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics. *Genet Med.* 2016;18(10):1056-65.

[2] Curtin SC and Abma JC. Centers for Disease Control and Prevention. 2010 Pregnancy Rates Among U.S. Women.

[3] American Pregnancy Association http://americanpregnancy.org/prenatal-testing/amniocentesis/.

[4] American College of Genetics and Genomics Final Position Statement on NIPS, October 2016. http://www.acmg.net/PDFLibrary/Fetal-Aneuploidy-Noninvasive-Prenatal-Screening-Update.pdf.

This evolution of professional guidelines is largely due to the plethora of published, peer-reviewed literature that has clearly established that NIPS is a superior screening method compared to conventional serum screening, regardless of maternal age. Beyond professional associations, a 2016 survey showed that over 80% of physicians would utilize NIPS if covered by insurance.[5] Further, important advocacy organizations, including the March of Dimes, the Down Syndrome Diagnosis Network, the Focus Foundation, Sidelines, and others, support access to NIPS for all women.

Pregnant women join the ranks of the disenfranchised when access is denied for genetic testing that is defined by leading organizations such as ACOG and ACMG to be part of routine prenatal care, regardless of age or other risk factors. If women request screening for common genetic conditions, they should have access to the test that provides the most sensitive risk assessment with the lowest false positive rate — NIPS — eliminating needless anxiety, avoiding unnecessary pregnancy loss, and improving the standard of care.

In cases where NIPS testing returns positive results, laboratory genetic counselors often relay the results and information to ordering physicians. At the request of the physician, the clinical genetic counselors are available to speak with patients about both positive and negative NIPS results to provide an understanding of the test result and the options for appropriate follow-up testing for diagnostic purposes. Definitive diagnostic testing is always recommended for positive NIPS results.

***FDA's Intervention Would Harm Patient Access, Undermine Efforts to Advance Comprehensive Diagnostic Reform***

The report's recommendation that FDA selectively regulate genetic tests and additional Laboratory Developed Tests (LDTs) as devices would significantly undermine patient access to innovative clinical tests and diagnostics. Moreover, there is a pending legislative initiative called the Verifying Accurate Leading-edge IVCT Development Act, "VALID", that is intended to regulate, in vitro diagnostics including laboratory tests, and therefore, requesting the FDA to begin to institute action apart from the VALID process is counter-productive and not complementary to the joint efforts of all the stakeholders who have been working on VALID, including labs and the FDA.

The Centers for Medicare and Medicaid Services (CMS) regulates all laboratory testing services through the Clinical Laboratory Improvement Amendments (CLIA), which includes strict standards to ensure quality. In recent years, FDA has made efforts to impose additional regulations on clinical laboratory services, using its medical device authority. As we have consistently stated, LDTs are not devices but are distinct services that require their own regulatory framework and should not be forced into existing and conflicting regulatory frameworks that are not designed specifically for diagnostics. To maintain patient access and incentivize the continued development of novel tests, we cannot regulate these tools with a one-size-fits-all approach that was designed for therapeutic products, and this is a key difference between the Council's proposal and the VALID solution.

Over the past several years, Congressional leaders have advanced legislative proposals in collaboration with stakeholders and the Obama and Trump Administrations, beginning with the Diagnostic Accuracy and Innovation Act (DAIA), and more recently, VALID. ACLA and our members are committed to working with Congress to advance comprehensive diagnostic reform to ensure high-quality, accurate, reliable tests are reaching the patients who need them, and that new regulations support future innovations for both patients and providers. To ensure the new regulatory framework has maximum

[5] Brewer J, Demers L, and T Musci. Survey of US obstetrician opinions regarding NIPS use in general practice: implementation and barriers. The Journal of Maternal-Fetal & Neonatal Medicine. DOI: 10.1080/14767058.2016.1225035.

and lasting benefits to patients, it is crucial that this reform go through Congress, rather than FDA and clearly, this solution is where efforts should be focused.

### *Value and Ethics of Genetic Counseling Profession*

Clinical labs are proud of our role in expanding patient and physician access to accredited genetic counselors as part of our effort to deliver a new era of personalized medicine. Responding to demand for these valuable genetic testing tools, the field of genetic counseling has grown significantly in recent decades. In 1993, the number of Certified Genetic Counselors (CGC) was just 495. Today there are more than 5,000.[6]

Genetic counselors help physicians, patients, and their families understand genetic risks, disorders and complex test results. These highly trained professionals work hand-in-hand with a patient's health care team to empower informed clinical decision-making. Although we understand the Council's concern regarding more focused disability training for genetic counselors, they fulfill a much broader educational function in helping to guide and educate health care practitioners and patients regarding all choices available to them. For example, when genetic counselors are asked to discuss Down syndrome, the counselors provide information on the natural history of the condition, associated pregnancy risks, genetic etiology, and risk of recurrence. They also provide written information about Down syndrome, and include a list of resources for patients including the National Down Syndrome Society, among others.

The Council's claim that these genetic counselors have conflicted interests ignores both the rigorous training and ethics of the profession. In reality, these individuals are obligated to follow a strict code of ethics that prohibits any entity from influencing their professional practice in a way that is not in the best interest of patients.[7] To be considered for certification, candidates must have a Master's degree in Genetic Counseling from a program accredited by the Accreditation Council for Genetic Counseling (ACGC) and also pass a formal exam. Failure to adhere to ethical requirements can result in loss of certification, and loss of certification can result in loss of employment for genetic counselors. Additionally, ethical health care providers and genetic counselors act in the best interests of patients, however, where fraudulent abuses may occur, genetic counselors and the laboratories that employ them are subject to federal and state fraud and abuse laws, including the False Claims Act, the Stark Law, and the Anti-Kickback Statute that prohibit overutilization based upon fraudulent activities. Finally, as noted above, clinical laboratories adhere to federal standards under CMS and CLIA related to laboratory services, as well as relevant state regulations and the standards of accreditation entities such as the College of American Pathologists (CAP).

With regard to NIPS testing specifically, we recognize that the decision to move forward with this screening is a highly personal decision for many women, which is why the National Society of Genetic Counselors (NSGC) offers cultural education so that counselors can appropriately incorporate discussion of religion and spirituality in genetic counseling sessions.[8] The NSGC Code of Ethics also mandates that all counselors "respect their clients' beliefs, inclinations, circumstances, feelings, family relationships, sexual orientation, religion, gender identity, and cultural traditions."[9] To the extent that further disability, cultural or social awareness training could be helpful that may not be provided by employers, such recommendations should be directed, more appropriately, toward the university degree programs and board certification authorities for genetic counselors rather than the laboratories that employ them afterward. We, however, do support the concept that training during all aspects of

[6] *See* National Society for Genetic Counselors Background, *available at* https://www.nsgc.org/p/cm/ld/fid=612
[7] *See* National Society for Genetic Counselors Code of Ethics, *available at* https://www.nsgc.org/p/cm/ld/fid=12
[8] *See* National Society for Genetic Counselors Genetic Counseling Cultural Competence Toolkit, *available at*
[9] *See* National Society for Genetic Counselors Code of Ethics, *available at* https://www.nsgc.org/p/cm/ld/fid=12

person's professional career is a positive and various post-graduate, educational opportunities such as continuing education requirements should be fully supported and these type initiatives could provide such opportunities without burdensome regulatory interventions.

***Conclusion***

To enable a new regulatory framework to have maximum and lasting benefits to patients, we cannot afford for FDA to undermine ongoing Congressional efforts to advance comprehensive diagnostic reform. Furthermore, arbitrarily imposing an additional layer of FDA device regulations — designed for manufactured therapeutic products rather than laboratory testing services — would be an injustice to the nearly 700,000 men and women working in the clinical laboratory industry in the U.S., whose driving purpose is to deliver quality care for their patients.

We appreciate your attention and consideration and look forward to the opportunity to discuss these issues at your earliest convenience.

In the meantime, should you have any questions, please feel free to reach me at 202-637-9466 or jkhani@acla.com.

Sincerely,

Julie Khani
President