

April 30, 2009

**VIA FACSIMILE And FEDEX**

Honorable Kathleen Sebelius  
Secretary, Department of Health and Human Services  
Hubert H. Humphrey Building  
200 Independence Avenue, S.W.  
Washington, D.C. 20201

**Re: Oversight of Advanced Diagnostic Tests**

Dear Secretary Sebelius:

Congratulations on your appointment as Secretary of Health and Human Services! What an amazing opportunity to help shape our healthcare system to meet the challenges of the 21<sup>st</sup> century.

One of the keys to achieving your goals and meeting these challenges will be taking full advantage of the prospects offered by personalized medicine—targeting therapies to individual patients to maximize their effective, safe, and efficient use. At the core of personalized medicine is advanced diagnostic testing that improves a physician’s ability to assess whether an individual patient is or is not likely to benefit from treatments for his or her disease or condition. Advanced diagnostic testing is becoming the standard of care for many diseases.

Accurate, reliable, and timely advanced diagnostics offer enormous promise, but poor quality testing can harm patients and waste scarce resources. Therefore, it is critical that regulatory oversight of these innovations (and innovators) strike the right balance between assuring patient safety and embracing policies that encourage the incorporation of rapidly advancing scientific methods and knowledge. To achieve this objective, a new regulatory framework is needed. Additionally, it is essential that new regulatory oversight policies be clearly stated and publicly vetted before they are implemented. To facilitate achieving these objectives for regulatory oversight, the undersigned individuals and groups recommend that you strive to achieve the following goals:

- Oversight of all advanced diagnostics (including *in vitro* diagnostic test kits and laboratory-developed tests [LDTs]) should be risk-based and built upon an expanding base of advanced scientific capacities within the Department of Health and Human Services (HHS) and its agencies and in collaboration with extramural experts to augment HHS’s capabilities as needed. Food and Drug Administration (FDA) regulation of advanced diagnostic LDTs can be implemented—and can strike the essential regulatory balance described above—only through formal notice-and-comment rulemaking.
- A registry should be developed and maintained that includes the name of the laboratory performing a specific test, the name of the laboratory or manufacturer that developed the test, and information to support claims about the analytical validity and clinical validity of that specific test or test method. Submission of information to this registry should be mandatory for all advanced diagnostic assays.
- Oversight of clinical laboratory quality systems by the CLIA program should be strengthened to assure that the information provided by advanced diagnostic testing is accurate, reliable and

timely. FDA and the Centers for Medicare & Medicaid Services (CMS) should avoid unnecessary duplication in oversight and reconcile any conflicts in regulation between the medical device rules and regulations under CLIA.

In 2007, Secretary Leavitt charged his Advisory Committee on Genetics, Health and Society to make recommendations for appropriate oversight of advanced diagnostic testing. The Committee engaged in vigorous debate and generated many thoughtful recommendations. We believe that now is a critical time to move to the next stage—to create and implement a reasonable and responsible regulatory framework for advanced diagnostic tests. The framework must put patients first, be grounded in science, appropriately incentivize innovation, and be fully consistent with established statutes.

The undersigned organizations represent the leading advocates for the advancement of personalized medicine and a broad spectrum of stakeholders with respect to these issues: the innovative laboratories that develop and offer advanced diagnostics, the physicians who utilize them in practice, the investors who believe in the promise of personalized medicine, and the patients who need more informed and targeted medical care. These groups, all representing different perspectives and with different priorities, have come together in a collaborative spirit and are committed to working with you to create this new system for advanced diagnostics and to see that it gets up and running in a timely manner.

We look forward to meeting with you to discuss this important opportunity in personalized medicine. Please contact Gail Javitt at 202.663.5972 (gjavitt1@jhu.edu) or Paul Radensky at 202.756.8794 (pradensky@mwe.com) if you have any questions or would like to discuss in advance of our meeting with you. We all share the responsibility to “get personalized medicine right.”

Sincerely yours,

5 Degrees Biosciences, Inc.  
Adult Polyglucosan Body Disease Research Foundation  
Alpha-1 Advocacy Alliance  
American Hemochromatosis Society, Inc.  
American Autoimmune Related Diseases Association, Inc.  
Amyloidosis Support Groups, Inc.  
Androgen Insensitivity Syndrome Support Group  
APS (Antiphospholipid Antibody Syndrome) Foundation of America, Inc.  
Ara Parseghian Medical Research Foundation  
Association of Gastrointestinal Motility Disorders, Inc. (AGMD)  
Axial Biotech, Inc.  
Biodesix, Inc.  
Bonei Olam, Inc.  
Brazilian Genetic Alliance  
Breast Cancer Network Of Strength  
Burrill & Company  
CADASIL (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy) Foundation  
Canadian Multiple Endocrine Neoplasia Society, Inc.

CardioDx, Inc.  
Cedar Associates LLC  
CFC (Cardio-Facio-Cutaneous Syndrome) International  
Children's Sickle Cell Foundation, Inc.  
Chromosome Disorder Outreach, Inc.  
Citizens for Quality Sickle Cell Care, Inc.  
Claire Altman Heine Foundation, Inc.  
Coalition For 21<sup>st</sup> Century Medicine  
Congenital Adrenal Hyperplasia Research Education and Support (CARES) Foundation, Inc.  
Cooley's Anemia Foundation, Inc.  
Crescendo Bioscience, Inc.  
Cutaneous Lymphoma Foundation  
Cutis Laxa Internationale  
Detroit Medical Reserve Corps  
Duke Institute for Genome Sciences & Policy Center for Genome Ethics, Law & Policy  
Expression Analysis, Inc.  
FORCE: Facing Our Risk of Cancer Empowered, Inc.  
Foundation for Ichthyosis & Related Skin Types, Inc.  
Foundation for Nager and Miller Syndromes  
Foundation for Prader-Willi Research  
FRAXA Research Foundation  
Geneforum  
Generation Health, Inc.  
Genetic Alliance  
Genetics & Public Policy Center at Johns Hopkins University  
Genomas, Inc.  
Genomic Health, Inc.  
Hereditary Hemorrhagic Telangiectasia Foundation International, Inc.  
Hereditary Spherocytosis Society  
Highland Capital Partners, LLC  
Histiocytosis Association of America  
Huntington's Disease Society of America, Inc.  
In Need Of Diagnosis, Inc. (INOD)  
Incontinentia Pigmenti International Foundation  
International Mosaic Down Syndrome Association  
Kids With Heart National Association for Children's Heart Disorders, Inc.  
Kleiner Perkins Caulfield, & Byers  
Leomics Associates, Inc.  
Maryland Patient Advocacy Group  
MDV-Mohr Davidow Ventures

MLD (Metachromatic Leukodystrophy) Foundation  
Monogram Biosciences, Inc.  
MPD Support- Myeloproliferative Disease Support and Daily Email Digest  
The Myelin Project  
Nanogen, Inc.  
National Foundation for Ectodermal Dysplasias  
National Fragile X Foundation  
National Hemophilia Foundation  
National Hispanic Medical Association  
National Marfan Foundation  
National Venture Capital Association  
National Tay-Sachs & Allied Diseases Association, Inc.  
Neurodegeneration with Brain Iron Accumulation (NBIA) Disorders Association  
New Leaf Venture Partners  
Northern Nevada Genetic Counseling  
Omicia, Inc.  
PKS Kids (Pallister-Killian Mosaic Syndrome)  
Parent Project Muscular Dystrophy  
Pediatric Adolescent Gastroesophageal Reflux Association  
Periodic Paralysis International/HKPP Listserv  
Perlegen Sciences, Inc.  
Personalized Medicine Partners, LLC  
Population Diagnostics, Inc.  
Prader-Willi Syndrome Association (USA)  
PRISMS (Parents and Researchers Interested in Smith-Magenis Syndrome)  
The Progeria Research Foundation, Inc.  
Pull-thru Network, Inc.  
PXE (Pseudoxanthoma Elasticum) International, Inc.  
Quebec Portal for Orphan Genetic Diseases  
RedPath Integrated Pathology, Inc.  
Research Advocacy Network  
Sarcoma Foundation of America, Inc.  
Science Futures LLC  
Sickle Cell Disease Association of America, Inc.  
Sirius Genomics, Inc.  
SomaLogic, Inc.  
Spina Bifida Association  
Stennis Foundation for the Leukodystrophies  
Sudden Arrhythmia Death Syndromes (SADS) Foundation  
Target Discovery, Inc.

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TcLand Expression SA  
Technic Solutions, LLC  
Tethys Bioscience, Inc.  
Trimethylaminuria Foundation  
United Leukodystrophy Foundation  
Veracyte, Inc.  
Versant Venture Management LLC  
VitaPath Genetics, Inc.  
Von Hippel-Lindau (VHL) Family Alliance  
XDx, Inc. (Expression Diagnostics)