THE NATIONAL CENTER FOR PRENATAL AND POSTNATAL DOWN SYNDROME RESOURCES

Stephanie Meredith, M.A. and Harold Kleinert, Ed.D.
Evidence-based and medically-reviewed information for new and expectant parents learning about a diagnosis of Down syndrome is becoming increasingly critical as the new technology for prenatal screening for Down syndrome (Non-Invasive Prenatal Screening or NIPS) becomes more accurate and available — leaving many patients feeling overwhelmed and isolated without immediate support and information to understand what a diagnosis of Down syndrome actually means for individuals and families.

And for parents of newborn children with Down syndrome receiving a diagnosis at birth, accurate, balanced information is also very important!
LEARNING OBJECTIVES

1. Understand the current landscape of prenatal testing.

2. Learn about the purpose of the National Center for Prenatal and Postnatal Down Syndrome Resources and what scientifically accurate, up-to-date, and balanced materials the National Center provides for new and expectant parents. Specifically discuss the ACMG and NSGC guidelines.

3. Learn how to address the needs of diverse populations undergoing prenatal testing, including different cultures.

4. Review how LEND programs in other areas of the country are using National Center materials to serve their populations.
PRENATAL TESTING BASICS
SCREENING VS. DIAGNOSTIC

• Traditional Screening
• Ultrasounds
• New Non-invasive Prenatal Screening
• Aminocentesis
• CVS
PRENATAL TESTING BASICS

Difference between sensitivity and positive predictive value.

<table>
<thead>
<tr>
<th>PRENATAL SCREENING/TESTING OPTIONS FOR TRISOMY 21 AND OTHER FETAL ANEUPLOIDIES</th>
<th>NIPT</th>
<th>Serum Screening</th>
<th>CVS and Amniocentesis</th>
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| **When** | 10 weeks and over | 10-22 weeks | CVS: 10-13 weeks  
Amniocentesis: 15-16 weeks and over |
| **Intended population** | Validated in:  
Advanced maternal age  
Personal or family history of aneuploidy  
Abnormal serum screen  
Abnormal ultrasound | All women | Women at increased for:  
Chromosome abnormalities  
Known inherited disorders  
ONTD |
| **How performed** | Maternal blood | Maternal blood | Invasive procedure |
| **Risks** | None | None | Risk of miscarriage:  
CVS: 1/100-1/200  
Amniocentesis: 1/200 -1/500 |
| **T21 – detection rate** | T21 – >99% | T21 – 80-95% | T21 – 99.99% |
| **False-positive rate** | Less than 1% | 3-5% | Less than 1% |
| **Other aneuploidies** | T18 – 99%  
T13 – 79-92% (limited data) | T18 – 80-95%  
T13 – uncertain | T18 – 99.99%  
T13 – 99.99% |
| **Potential for other screening information** | Some sex chromosome abnormalities – uncertain  
ONTD (2nd trimester screening) – 90-95%  
Risk for poor pregnancy outcome – uncertain | Some sex chromosome abnormalities – uncertain  
ONTD (Amniocentesis) – >95% | Other numerical and structural chromosome abnormalities – 99.99%  
ONTD (Amniocentesis) – >95% |
| **Other genetic testing possible** | None | None | Known inherited disorders |
| **Turn around time** | Week or more | 2-3 days | Up to 2 weeks |
| **How reported** | Reported as positive/consistent with, negative, and in the case of one laboratory, a numerical risk assessment is provided. Some laboratories may provide a likelihood ratio with the negative result, allowing the clinician to revise an a priori risk for trisomy | Reported as a revised risk based on maternal age-related risk, results of serum screen, and nuchal translucency (if done) | Reported as a normal or abnormal karyotype |

Non-Invasive Prenatal Testing Fact Sheet created by NCHPEG/NSGC with a grant from Sequenom
NON-INVASIVE PRENATAL SCREENING

• Ariosa Diagnostics® Harmony Test™
• Sequenom CMM® Materniti21 Plus™ test
• Natera™ Panorama test™
• Verinata® Verifi® test
Column: Will America cull people with Down syndrome?

By Brian Skotko

Updated 11/4/2011 5:02 PM
Comment 97 | Recommend 0 | Tweet 136 | +1 2

In mid-October, pregnancy as we know it forever changed in America. The company Sequenom announced with a simple blood draw at 10 weeks of gestation, a pregnant woman can now learn with near 99% accuracy whether her fetus has Down syndrome.

As a physician in this news brings so “negative” result could be to avoid risks or chorionic villus long needle through to obtain a sample chromosomal these procedures.

Eventually, the

The end of Down syndrome

A controversial new test means disorder will become extinct from ‘eugenics,’ doctors say

By MAYRAV SAAR
Last Updated: 12:29 PM, November 13, 2011
Posted: 9:53 PM, November 12, 2011

A new, simple way to detect Down syndrome in a fetus means the condition will be virtually extinct — but not without a great deal of controversy, experts say.

Last month, San Diego-based Sequenom released a test that allows doctors to screen for the most prevalent type of Down syndrome with only a blood test from the mother. The screening is available in 20 cities and is expected to hit New York soon. Two other companies have plans to release similar tests next year.
THE DILEMMA

• Prenatal testing technology is developing rapidly.

• Women who are pregnant or who plan on becoming pregnant say they want balanced, up-to-date, and scientifically accurate information after a diagnosis that includes information about living with the condition and all pregnancy options.

• There is no federal legal requirement or funding for accurate and up-to-date information.

• Research indicates the majority of patients do not receive accurate and up-to-date information about Down syndrome.
THE NATIONAL CENTER FOR PRENATAL AND POSTNATAL DOWN SYNDROME RESOURCES

DOWNSYNDROMEDIAGNOSIS.ORG
• University of Kentucky’s Human Development Institute

• Brighter Tomorrows

• Lettercase

• Down Syndrome Pregnancy
REACHING OUT TO MEDICAL PROFESSIONALS
SUPPORTING EXPECTANT PARENTS
BRIGHTER TOMORROWS
BRIGHTERTOMORROWS.ORG

For prenatal and postnatal parents:

- **Free** online English and Spanish multi-media resource with information about testing, prenatal topics, and postnatal topics; videos; life glimpses; and free downloadable pamphlets reviewed by medical professionals to provide both prenatal and postnatal support

- Funded by the National Center on Birth Defects and Developmental Disabilities, through a cooperative agreement with the Association of University Centers on Disabilities

- Created in 2008 and updated in July 2012
Published Effectiveness Studies


LETTERCASE
LETTERCASE.ORG
For pregnant women before testing:

- NEW “Understanding Prenatal Screening and Testing for Chromosome Conditions” in English and Spanish

- Gives an overview of prenatal screening and testing (including NIPS/NIPT), states reasons why expectant parents might or might not want these tests, explains testing is optional, gives a brief summary of the different conditions that can be detected with prenatal screening, lists the national support organizations for each condition (sex chromosome and trisomy conditions and Spina Bifida, and shows photos of different conditions

- Reviewed by professional advisory committee: scholars, genetic counselors, an OB/GYN, a geneticist, educations experts, a pediatrician, legal experts, disability advocates, and patient support experts

- Available for pre-order throughout October ($1 for a minimum purchase of 50)
For expectant parents first receiving a diagnosis

- The Kennedy Foundation’s “Understanding a DS Diagnosis” and “Delivering a DS Diagnosis” available in print and as free digital downloads in English and Spanish. Over 35,000 copies distributed nationwide and used by over 100 local organizations, hospitals, public health departments, medical practices, and Ariosa.

- Created in 2008 and selected in 2009 as the NDSS/NDSC First Call gold standard

- Revised with assistance in 2010 from representatives of the American Congress of Obstetricians and Gynecologists (ACOG), the National Society of Genetic Counselors (NSGC), the American College of Medical Genetics and Genomics (ACMG), the National Down Syndrome Society (NDSS), and the National Down Syndrome Congress (NDSC)

- Endorsed by 45 scholars and physicians as “the most balanced material that healthcare providers should give to expectant parents following a prenatal diagnosis.”
Survey results from 41 medical providers:

- All who used the book said it was helpful with 80% saying it was "very helpful."

- "I think they are very informative, while also being neutral. A patient can feel like they are not receiving biased information."

- "The engaging pictures - beautiful children doing lots of normal things. The text - simple to understand, balanced, positive."

- "Although I LOVE the ADORABLE pictures of all of the kids with DS, I think this may be a little unfair for parents who are trying to make a choice between continuing and terminating a pregnancy. Maybe it would be reasonable to include a couple of pix of babies or kids who are "less cute," as well as some older adults. Maybe a child in the hospital after heart surgery or an older adult who clearly needs assistance with daily living activities."

- "I would definitely say more about first trimester screening and ultrasound. These are well established and non-invasive, as well as being far less expensive than cell free DNA screening. They also help detect other more serious defects and complications, which ultimately may be more important."
Website for expectant parents throughout pregnancy

- Includes the medically-reviewed books *Diagnosis to Delivery: A Pregnant Mother’s Guide to Down Syndrome* (2010) and *Your Loved One is Having a Baby with Down Syndrome* (2012), available as free downloads and through Woodbine House Publishing Company

- Website created in late 2010 includes constantly updated online resources and information for expectant parents, local Down syndrome organizations, medical providers, friends and family, and the community.

- Site viewed by nearly 140,000 visitors worldwide and book downloaded by nearly 2500 expectant parents, medical providers, and others.
PREGNANCY TIMELINE: LAYERS OF SUPPORT
THE NATIONAL CENTER
DOWNSYNDROMEDIAGNOSIS.ORG

National Center materials cited in:

• Genetics in Medicine

• American Journal of Medical Genetics

• American Journal of Obstetrics and Gynecology

• NSGC Guidelines for Delivering a Diagnosis of DS

• Journal of Midwifery and Women's Health

• Journal of Genetic Counseling

• Obstetrics and Gynecology
National Center program initiatives and distribution plans:

- Offer presentations at national medical/genetics conferences such as NBDPN, ACMG, NSGC, and Down Syndrome leadership annual meetings.

- Participate in national studies such as the Duke Down Syndrome Prenatal Diagnosis study.

- Support more than 65 local Down syndrome organizations in 36 states.

- Manage targeted distributions to medical professionals. More than 35,000 copies of the Kennedy Foundation’s “Understanding a Down Syndrome Diagnosis distributed in 5 years including distributions directly to all members of NSGC, ACMG, and genetic counseling students. Featured in ACMG and NSGC newsletters, guidelines, and on websites.

- Negotiate distributions with biotech companies. Materials distributed by Ariosa Diagnostics at prenatal medical conferences in 2013.

- Direct online and print distribution to thousands of medical providers, hospitals, medical offices, and expectant parents annually.
Materials from the National Center have been a focal point for state laws in Massachusetts and Kentucky

- **Massachusetts** *An Act Relative to Down Syndrome Genetic Test Results (H3825)* June 22, 2012: “Medical professionals are required to give parents who receive a prenatal or postnatal diagnosis "up-to-date, evidence-based, written information about Down syndrome that has been reviewed by medical experts and national Down syndrome organizations." The written information provided must include "physical, developmental, educational, and psychosocial outcomes, life expectancy, clinical course, and intellectual and functional development and treatment options." All of this information must be culturally and linguistically appropriate.

- MA Department of Public Health purchases 2,000 copies of the Lettercase booklet in 2013

- **Kentucky** *The Down Syndrome Information Bill* Senate Bill 34: “Up-to-date, evidence-based, written information about Down syndrome that has been reviewed by medical experts and Down syndrome organizations and includes information on physical, developmental, educational, and psychosocial outcomes, life expectancy, clinical course, intellectual and functional development, and treatment options;”
PROFESSIONAL POLICIES

• The American College of Medical Genetics and Genomics (ACMG) policy statement (2013) indicates that patients should be provided accurate, up-to-date, and balanced information about Down syndrome upon receiving a diagnosis. The statement specifically includes Lettercase, Brighter Tomorrows, and the American Academy of Pediatrics “Health-Care Supervision for Children with Down Syndrome” as recommended patient resources.

• The NSGC Practice Guidelines for Communicating a Diagnosis of Down Syndrome (2011) recommends Lettercase, Brighter Tomorrows, and Down Syndrome Pregnancy.

“They’re penetrating the bureaucracy!”

- The Incredibles
“Expectant parents deserve accurate and up-to-date information about Down syndrome. We endorse “Understanding a Down Syndrome Diagnosis” as the most balanced material that healthcare providers should provide to expectant parents following a prenatal diagnosis. This material was uniquely prepared with assistance from The Down Syndrome Consensus Group, including representatives of the American Congress of Obstetricians and Gynecologists, American College of Medical Genetics and Genomics, the National Society of Genetic Counselors, the National Down Syndrome Congress, and the National Down Syndrome Society.”
SUPPORTING EXPECTANT PARENTS

“When I got the news that my baby girl had Down syndrome, my world turned upside down. Downs syndromepregnancy.org launched within weeks of getting Ellie’s diagnosis. I was able to share the website and book with family members and close friends; connect with other women; learn more about Down syndrome; and find hope from experienced parents who talked me through both of my daughter’s major surgeries. I cannot fathom how difficult my pregnancy would have been without the voices of other parents providing hope.

Megan Landmeier (www.mystubbornmiss.com)
SUPPORTING EXPECTANT PARENTS

Dyan, the mother of a 6 month-old with Down syndrome, “These materials were a lifeline to me during my pregnancy ... It was a bright spot for me in an otherwise dark and scary time.”
LEND PROGRAMS AND MEDICAL OUTREACH INITIATIVES

• Oklahoma LEND Program

The Down Syndrome Association of Central Oklahoma has worked with the University of Oklahoma Health Sciences Center to provide prenatal materials to patients and share costs.

• Nevada Center for Excellence in Disabilities

The Nevada UCEDD assigned a Family Studies Student Intern to research all medical professionals in the area delivering a diagnosis of Down syndrome and delivered books directly to them. Worked with Down Syndrome Network of Northern Nevada.

• More Ideas? Sharing resources on websites, sharing resources with genetic counseling/genetics students, ...
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