Identifying Needs, Implementing Services in the Indiana Plain Community

Chris Roberson, JD, MPH  
*Director of Compliance & Community Programs*  
The Indiana Hemophilia & Thrombosis Center  
*Board of Directors*  
The Community Health Clinic

Rebecca Evans, LCGC  
*Genetic Counselor*  
The Indiana Hemophilia & Thrombosis Center  
The Community Health Clinic

*A Place for Our Special Children*
The Indiana Plain People

- **Indiana**
  - 47,235 Amish Individuals
  - 22 settlements

- **3rd largest Amish settlement located in northern IN**
  - Elkhart and LaGrange counties contain 21,560 Amish individuals

- **Mennonites in the area as well**
HTC Comprehensive Model of Care

- Non-profit/mission driven
- **Multidisciplinary**
  - Physician, PA/NP, Nurse(s), Physical Therapy, Social Workers, Dietitian, Dental Hygiene, Genetic Counseling, Risk Reduction, Research, Career Counseling
- Family/patient centered
- Part of national network of 140 centers
- Public Health Service pharmacy funded
<table>
<thead>
<tr>
<th>Diagnosis</th>
<th># Amish patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Factor IX deficiency</td>
<td>48</td>
</tr>
<tr>
<td>Factor VIII deficiency</td>
<td>2</td>
</tr>
<tr>
<td>VWD Type 1</td>
<td>15</td>
</tr>
<tr>
<td>VWD Type 2M</td>
<td>74</td>
</tr>
<tr>
<td>PAI-1 deficiency</td>
<td>11</td>
</tr>
<tr>
<td>Factor IX low level carrier</td>
<td>22</td>
</tr>
<tr>
<td>Factor IX non-low level carrier</td>
<td>50</td>
</tr>
<tr>
<td>Factor VIII low level carrier</td>
<td>1</td>
</tr>
<tr>
<td>Factor VIII non-low level carrier</td>
<td>8</td>
</tr>
</tbody>
</table>
- **FIX deficiency (XL)**
  - 31008 C>T
- **FVIII deficiency (XL)**
  - Intron 22 inversion
- **VWD 2M (AD)**
  - 4120 C>T
- **VWD 1 (AD)**
  - Mutation unknown
- **PAI-1 deficiency (AR)**
  - 2bp insertion (TA) at 3’ end of exon 4 resulting in a frameshift and new stop codon
Clotting factor concentrate utilized by the Amish in Indiana
The Community Dental Clinic
The “Amish CDC”
The Community Health Clinic
What has the CHC done to date?

2009

- Established as a non-profit 501(c)(3) organization
- Collaborating with Dr. Morton in Pennsylvania & Dr. Wang in Ohio
- Wrote a Business Plan
- Formed board of directors and began meeting
- Fundraising: Community Auction
- Bought land for permanent site

2010

- Physician recruitment
- Private foundation grant writing
- Fundraising from private donors
- Newsletter
What has the CHC done to date?

2011
- Physician recruitment
- Renovated temporary space in the basement of Dr. Egli’s office
- Fundraising
  - 2011 Dutch Dinner
- Financial Counseling with Melvin Miller
- Initiated Genetic Survey of the community

2012
- Launched bill counseling/negotiating service
- Abstracts presented at ACMG/SIMD
- Survey Implementation
- Launched the www.indianachc.org website
The need for bill negotiating services
2012-13

- Forged relationship with University of Michigan & Parkview Health Systems

- State Department of Health Grants
  - NBS Follow-up
  - MCH – Genetics Medical Home

- Hired Practice Manager, Nurse Practitioner, 2nd Physician
  - Recruiting Nurse, Dietitian, Receptionist/MA
Plans in 2013

- Continue to prepare for the opening of the CHC
- CHC Survey of all households to identify families needing our services
- **Fundraising**
  - 2013 Dutch Dinner
  - Apply for grant funding
  - Plan for a 2014 Community Auction
- **Most importantly, open the clinic to start seeing and treating patients**
- **Build an education resource room**
Future plans beyond 2013

- Start a CHC research program
- Expand lab services offered at the CHC
- Implement an in-house pharmacy
  - Will include some natural supplements & pharmacist who understands both medications and supplements and their interactions
- Build a permanent location
- Expand to primary care
Preliminary results of the CHC’s community-wide genetic survey
The CHC Survey

- **Purpose of survey**
  - Catalogue and determine prevalence of genetic disorders
  - Identify needs/allocate resources

- **Process of the survey**
  - Pilot phase, revisions, full phase

---

Dear Community Member,

We would like to improve healthcare services for your community. As board members of the Community Health Clinic, we would like to identify any special needs that your community has related to medical conditions that can be inherited or passed onto children through genes. These conditions may cause a variety of medical problems, some of which are very serious and may not be prevented if diagnosed and treated early enough. Information from this survey will help us create a clinic equipped to treat these medical conditions that affect your community.

Please fill out this survey and return it in the postage-paid return envelope mailed to your Community Health Clinic. Complete it as soon as possible. Even if no one in your household has any medical conditions that you are concerned about, you should still complete the survey. Please know that all your answers will remain confidential.

This survey is voluntary. Your decision whether or not to participate will not affect your ability to get quality care at the Community Health Clinic. Thank you, in advance, for your help to make the Community Health Clinic a place that serves your needs.

For questions about the survey:

Rebecca Davis
Genetic Counselor
Indiana Hemophilia & Thrombosis Center
Toll Free: 877-274-6807
Direct Line: 317-274-4351, ext. 222
Email: rebecca@HTC.org

---

I. Family status

1. Are you married? □ Yes □ No
2. Do you have any children? □ Yes □ No
   a. If yes, how many (including deceased children)? ______

II. Diagnosed Genetic Conditions

The next questions are about diagnosed medical conditions in your family that can be inherited or passed down to children through genes. Genes are like instructions that influence how our bodies grow, look, and work and also how we learn. Genetic conditions are caused by a change or difference in a person’s genes that causes them to not work correctly. Sometimes these changes, or differences, are inherited or passed down from a parent, and sometimes they happen for the first time in the child. Genetic conditions usually cause a child to have special needs (low learning or intellectual disability, seizures or convulsions, birth defects, cerebral palsy, etc.). When a baby is born with a genetic condition, signs are often present at birth or shortly after. However, some people may not show signs until later in life.

3. Have you, your husband or wife, or your living children ever been diagnosed by a doctor with any of the following:
   a. Genetic/inherited/metabolic disorder? □ Yes (List below) □ No (ex. PKU, congenital adrenal hyperplasia, muscular dystrophy, etc.)
   b. Birth defect? □ Yes (List below) □ No (ex. cleft lip or heart defect)
   c. Autism and/or autism spectrum disorder? □ Yes (List below) □ No

---

<table>
<thead>
<tr>
<th>Full Name</th>
<th>Date of Birth (MM/DD/YY)</th>
<th>Relationship</th>
<th>Lives in household?</th>
<th>Age at Diagnosis</th>
<th>Diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Joe Smith</td>
<td>02/01/19</td>
<td>Son</td>
<td>Yes</td>
<td>1 month</td>
<td>PKU</td>
</tr>
</tbody>
</table>
Progress

- **200 Amish church districts engaged**
  - ~5,052 households
  - ~23,744 individuals

- **141 districts returned some surveys**
  - 2,296 households
  - 11,199 individuals

- **Current response rate**
  - Households: 45%
  - Individuals: 47%
Progress

- **209 church districts in catchment area**
  - LaGrange, Elkhart, & Nappanee area
- **62 districts engaged to date**
  - ~1,157 households
  - ~5,400 - 8,400 individuals
- **28 districts returned some surveys**
  - 272 households
  - 1,351 individuals
- **Current response rate**
  - Households: 23.5%
  - Individuals: 16-25%
Survey Results To Date

No Special Needs, 10499, 94%

- Complex Condition, 165, 2%
- Birth Defect, 78, 1%
- Chromosome, 30, 0%
- Carrier, 28, 0%
- Metabolic, 41, 0%
- Other genetic, 132, 1%
- Mental health, 36, 0%
- Suspected, 190, 2%
<table>
<thead>
<tr>
<th>Condition</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Turner syndrome</td>
<td>1</td>
</tr>
<tr>
<td>Trichothiodystrophy</td>
<td>1</td>
</tr>
<tr>
<td>Ruvalcaba-Myhre syndrome</td>
<td>1</td>
</tr>
<tr>
<td>Propionic acidemia</td>
<td>1</td>
</tr>
<tr>
<td>PAI-1 clotting polymorphism</td>
<td>1</td>
</tr>
<tr>
<td>Nevus sebaceous syndrome</td>
<td>1</td>
</tr>
<tr>
<td>Neurofibromatosis</td>
<td>1</td>
</tr>
<tr>
<td>MCAD deficiency</td>
<td>1</td>
</tr>
<tr>
<td>Incontinentia pigmenti</td>
<td>1</td>
</tr>
<tr>
<td>Hemachromatosis</td>
<td>1</td>
</tr>
<tr>
<td>Gitelman syndrome</td>
<td>1</td>
</tr>
<tr>
<td>Crigler-Najjar syndrome</td>
<td>1</td>
</tr>
<tr>
<td>Cartilage hair hypoplasia</td>
<td>1</td>
</tr>
<tr>
<td>Arteriovenous malformation</td>
<td>1</td>
</tr>
<tr>
<td>Antithrombin (AT3) deficiency</td>
<td>1</td>
</tr>
<tr>
<td>Alpha 1-antitrypsin deficiency</td>
<td>1</td>
</tr>
<tr>
<td>22q13.3 deletion syndrome</td>
<td>1</td>
</tr>
<tr>
<td>1p36 deletion syndrome</td>
<td>1</td>
</tr>
<tr>
<td>MSUD</td>
<td>2</td>
</tr>
<tr>
<td>Methylmalonic acidemia</td>
<td>2</td>
</tr>
<tr>
<td>Hirschprungs</td>
<td>2</td>
</tr>
<tr>
<td>GM3 synthase deficiency</td>
<td>2</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>2</td>
</tr>
<tr>
<td>Factor V Leiden</td>
<td>2</td>
</tr>
<tr>
<td>Dilated cardiomyopathy</td>
<td>2</td>
</tr>
<tr>
<td>Congenital diaphragmatic hernia</td>
<td>2</td>
</tr>
<tr>
<td>Muscular dystrophy</td>
<td>3</td>
</tr>
<tr>
<td>Duchenne muscular dystrophy</td>
<td>3</td>
</tr>
<tr>
<td>Cystic hygroma</td>
<td>3</td>
</tr>
<tr>
<td>Club foot</td>
<td>3</td>
</tr>
<tr>
<td>Polycystic kidney disease</td>
<td>4</td>
</tr>
<tr>
<td>Neural tube defects</td>
<td>4</td>
</tr>
<tr>
<td>Severe combined immunodeficiency (RAG1)</td>
<td>5</td>
</tr>
<tr>
<td>16p11.2 microdeletion</td>
<td>5</td>
</tr>
<tr>
<td>Trisomy 18</td>
<td>6</td>
</tr>
<tr>
<td>Other aneuploidy</td>
<td>7</td>
</tr>
<tr>
<td>Cystic fibrosis</td>
<td>7</td>
</tr>
<tr>
<td>Hypospadias</td>
<td>8</td>
</tr>
<tr>
<td>Hypertrophic cardiomyopathy</td>
<td>8</td>
</tr>
<tr>
<td>Down syndrome</td>
<td>9</td>
</tr>
<tr>
<td>Congenital adrenal hyperplasia</td>
<td>14</td>
</tr>
<tr>
<td>Charcot Marie Tooth disease</td>
<td>14</td>
</tr>
<tr>
<td>Cleft lip +/- palate</td>
<td>15</td>
</tr>
<tr>
<td>Hemophilia</td>
<td>17</td>
</tr>
<tr>
<td>Congenital heart defect</td>
<td>19</td>
</tr>
<tr>
<td>PKU</td>
<td>31</td>
</tr>
<tr>
<td>Von Willebrand disease</td>
<td>36</td>
</tr>
</tbody>
</table>
Maple syrup urine disease 12
Zellweger syndrome 14
Spinal muscular atrophy I 5
Refsum disease 32
ITCH E3 ubiquitin ligase deficiency 12
Cystic fibrosis 5
Symptomatic epilepsy and skull dysplasia 15
GM3 synthase deficiency 24
Propionic acidemia 7
Methylmalonic acidemia 43
Duchenne muscular dystrophy 52
Charcot-Marie-Tooth disease 9
Severe combined immunodeficiency - RAG1 84
Severe combined immunodeficiency - ADA deficiency 93
Nonketotic hyperglycinemia 76
Dystonia 6
Phenylketonuria 222
Congenital adrenal hyperplasia 27
Cartilage-hair hypoplasia 1118
Limb-girdle muscular dystrophy 946

Deceased
Living