“To Cure Sometimes, To Relieve Often, To Comfort Always”

*DDC Clinic mission:* To enhance the quality of life for people with special needs caused by rare genetic disorders
DDC Clinic – Started with a Family Story

• An Amish family with four children suffering from a devastating condition tried to find the answer

• In 1998, five families traveled hundreds of miles to PA to see Dr. Holmes Morton

• A discussion about a new clinic was initiated on their way back
DDC Clinic –
Started with a Family Story

- A clinic was started with $50 collected in an Amish hat
DDC Clinic – History

• Inspired by Dr. Holmes Morton’s clinic in Lancaster, PA
• Community-based initiative for a dream of
  • A gathering place
  • A place of love, compassion, and caring where our children and families are respected
  • A place where people take time to listen and share
  • A place of faith and hope
In 1999, a 501(c)(3) non-profit organization started in Middlefield, Geauga County of Ohio
- Total population ~95,000, Amish ~14,000 (15%)
- 50% of developmental disabilities are from Amish
- One hour (but a world) away from world class healthcare

A collaboration of families, Amish communities, medical professionals, and research scientists
- Comprehensive patient care
- Patient-oriented translational research
- Community-based education
DDC Clinic – Mission and Vision

• **Our Mission**
  
  To enhance the quality of life for people with special needs caused by rare genetic disorders

• **Our Vision**
  
  DDC Clinic is a world-class medical home
Medical Home

- Medical home is **not** a building, house, hospital, or home healthcare service, but rather an approach to providing comprehensive primary care.
- “A medical home is defined as primary care that is accessible, continuous, comprehensive, family centered, coordinated, compassionate, and culturally effective.”

*The American Academy of Pediatrics (AAP)*
A Journey of Hope

Initial Visit

- 22%

Current Status

- 72%
DDC Clinic’s LEED Gold Facility Opened in 2009

LEED: Leadership in Energy & Environmental Design
DDC Clinic – A Unique Collaboration

- Serves about 720 patients from 32 States, Canada, Asia and Australia, with over 80 different diseases
- Over 80% of our patients are Amish or Mennonite
- No family is turned away based on their ability to pay
The New York Times

...“It may be too late for us, but if we can help others with this center, we’ll gladly do that” ...

From an Amish father with 4 special needs children (New York Times 6/20/2002)
Perspective in Medicine

To cure sometimes
To relieve often
To comfort always

(Dr. Edward Livingston Trudeau, 1848-1915)
Genomic Medicine

To cure sometimes
To relieve often
To comfort always
Genomic Medicine

To cure sometimes
Glucose/Galactose Malabsorption

- Autosomal recessive disorder
- Less than 100 patients reported worldwide
- Manifestations: Severe diarrhea, dehydration and malnutrition
- A potentially lethal disease, but very treatable
- Treatment: carbohydrate-free or low-carbohydrate diet
- DNA testing with cord blood
Conner Teare’s Story
To relieve often

Many genetic conditions fall into this category

- Cohen syndrome
- TMCO1 defect syndrome
- Prolidase deficiency
- Ganglioside GM3 synthase deficiency
- SAMS association
Genomic Medicine

To comfort always

- Examples:
  - Chicken breast disease
  - Cockayne syndrome
  - MOPD (Microcephalic Osteodysplastic Primordial Dwarfism) Type I
Translational Genomic Medicine in Plain Populations

Where do we stand and where are we going?

To cure sometimes
To relieve often
To comfort always

- Diagnosis – early, quick, reliable, affordable
- Research – patient-oriented, translational
- Treatment – new directions, novel approaches
Molecular Diagnostics Laboratory

- A CLIA certified genetic lab to provide high complexity clinical tests
- Only one of few labs focusing on plain communities in the country
- An affordable service compared with those in major medical centers
- Offering over 40 targeted mutations tests
Clinical Use of New Technologies

- Chromosomal microarray (CMA)
  - CytoScan HD – with more than 2.6 million SNPs
- Next-generation sequencing
  - Autism sequencing panel – 101 genes
  - Cardiomyopathy panel – 46 genes
  - Mendelian inherited disease panel – 552 genes
- Plain population panel – over a hundred of diseases
New Treatments and Clinical Trials

- Ganglioside GM3 synthase deficiency
- Neurodegeneration with brain iron accumulation 1 (NBIA1)
- Pyruvate kinase deficiency
- Phenylketonuria (PKU)
- SAMS association
Genomic Medicine – in 20 Years?

To cure sometimes
To relieve often
To comfort always
Dedicated to

- Plain communities – we have the privilege to work with
- Countless supporters – you make it happen
- The children and families we serve – you are our inspiration everyday