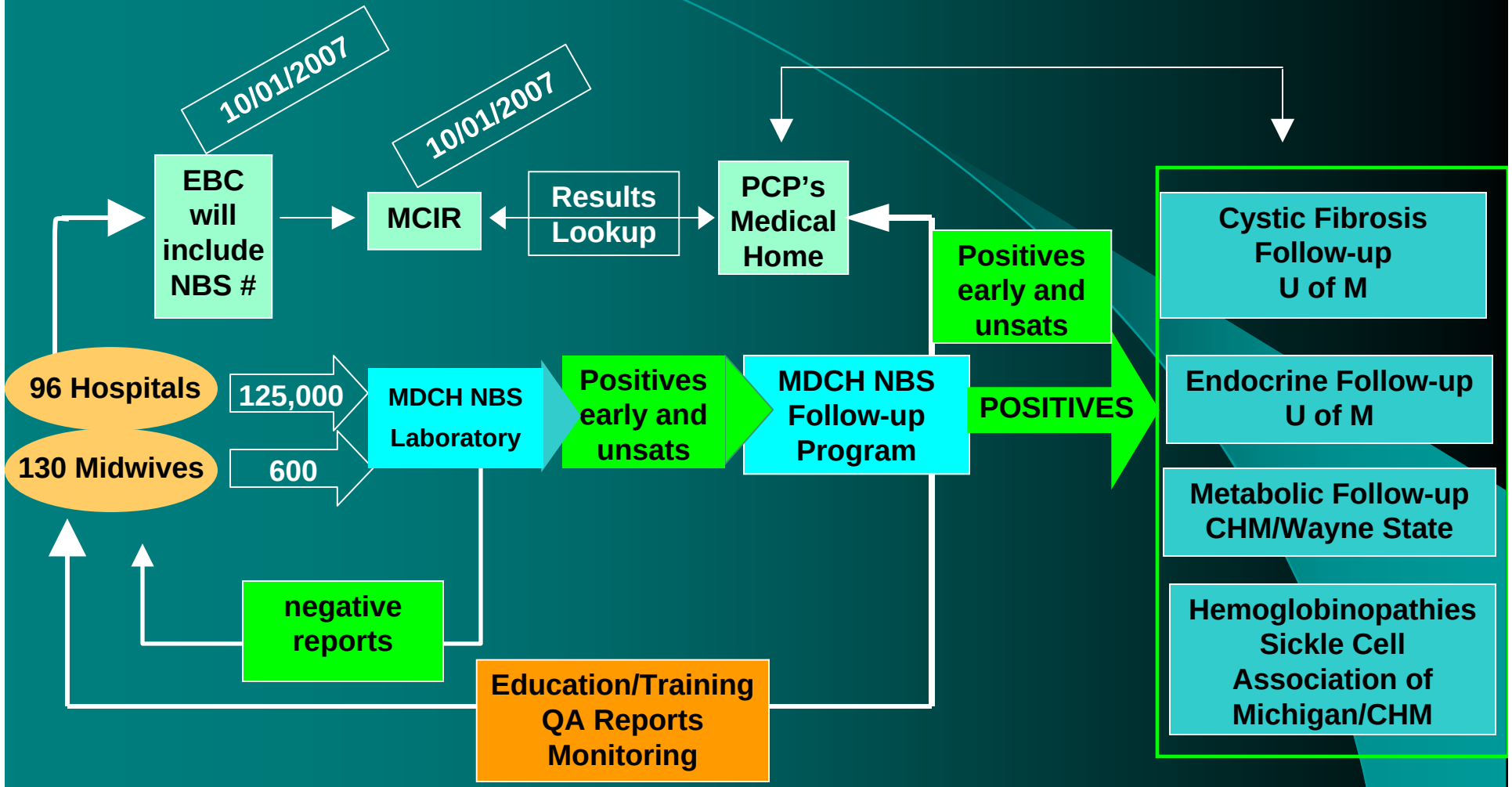
Michigan Newborn Screening

09-25-2007

MICHIGAN NEWBORN SCREENING OVERVIEW



NBS LABORATORY

Average 500 specimens each day

ENDOCRINE

- **17-OHP-CAH**
- **TSH-CH**

OTHER DISORDERS

- **Galactosemia**
- **Biotinidase Deficiency**
- **Cystic Fibrosis**

MS/MS

- **Amino Acid Disorders 12**
- **Fatty Acid Oxidation Disorders 13**
- **Organic Acid Disorders 15**

HEMOGLOBINS

- **HPLC/IEF**
- **A,F,S,C,D,E, Bart's**
- **SS, Sβ-thal, SC, SD, SE, HbC, HbE**

Disorders Identified via Newborn Screening, Michigan Residents, 1965-2006

Type of Disorder Classification (Year Screening Began)	Cases in 2006 (N)	Cases Through 2006 (N)	Cumulative Detection Rate*
Galactosemia (1985)	11	116	1: 24,850
Biotinidase Deficiencies (1987)	20	148	1: 16,928
Amino Acid Disorders (1965)	16	590	1: 9,901
Organic Acid Disorders (2005)	8	10	1: 12,752
Fatty Acid Oxidation Disorders (2003)	11	39	1: 9,951
Congenital Hypothyroidism (1987)	59	1,394	1: 1,870
Congenital Adrenal Hyperplasias (1993)	3	103	1: 16,819
Hemoglobinopathies (1987)	63	1,336	1: 1,951

MDCH FOLLOW-UP

Follow-up of positive specimens = 3200
Follow-up of unsat and early = 5200

Education – Primarily Hospital staff
Pediatricians and Family Practice Physicians

Monitoring Hospitals and Midwives

Assure diagnosis and medical management
Contracts with U of M, CHM and Sickle Cell Association

Family Support Group

Medical Home and Transitioning to Adult Care

METABOLIC FOLLOW-UP

MDCH NBS Follow-up Program
2006
Referrals 738
Diagnosed 67

Grand Rapids Clinic
2007

Molecular Genetics Laboratory

Gerald Feldman, M.D.

- Galactosemia
- MCAD
- LCHAD
- CF
- Connexin
- Sickle Cell

WSU/CHM
CHMMC
Gerald Feldman, M.D.,PhD
Erawati Bawle, M.D.
Laura Martin, M.D.
David Stockton, M.D.
Peggy Rush. M.S., CGC
Allison Hart, N.P.
Tammy Kelly, R.N.
June Vertimiglia, R.D.
Heidi Edwards, R.D.

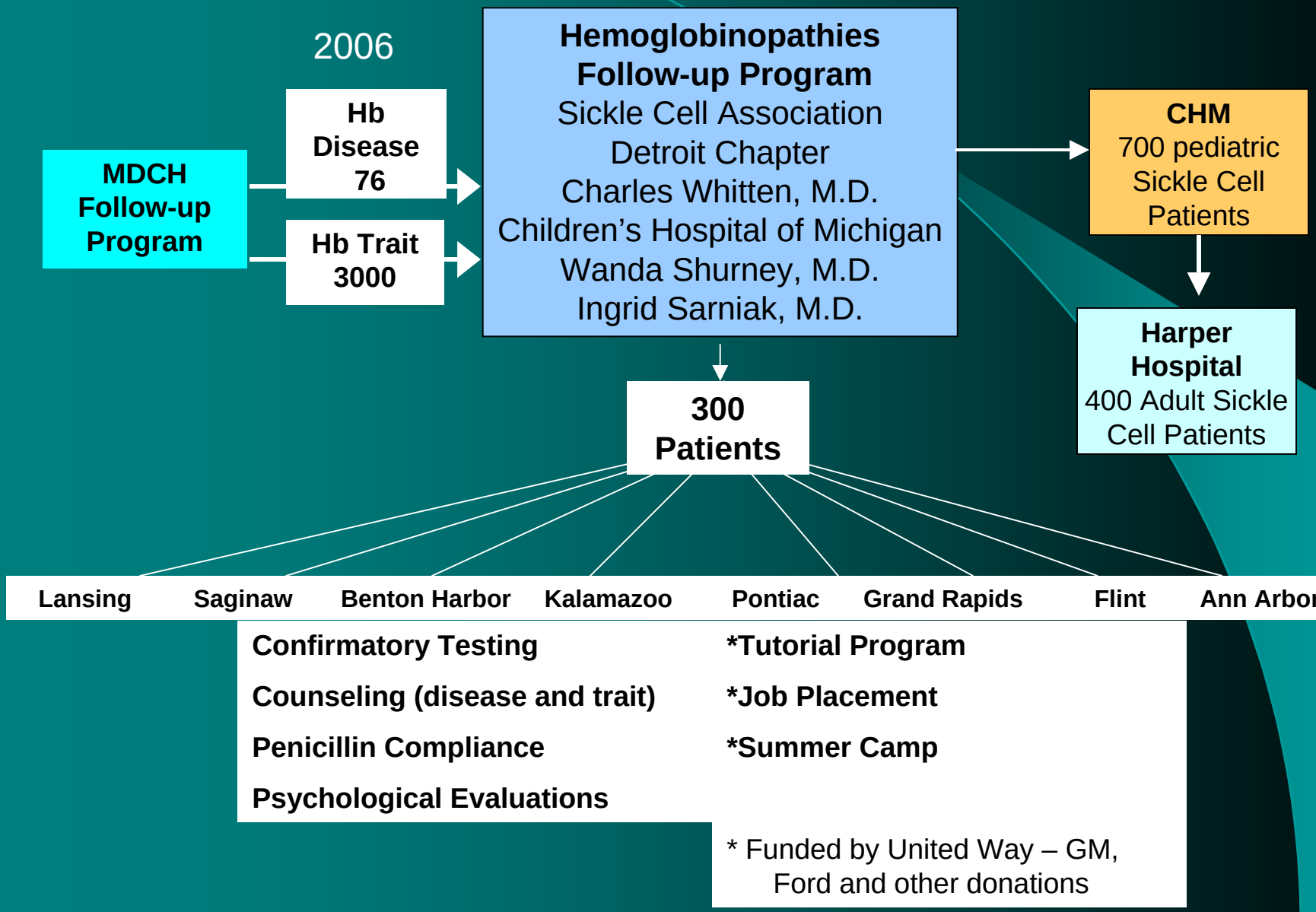
Total patients 903

Biochemical Genetics Laboratory
Bob Grier, PhD

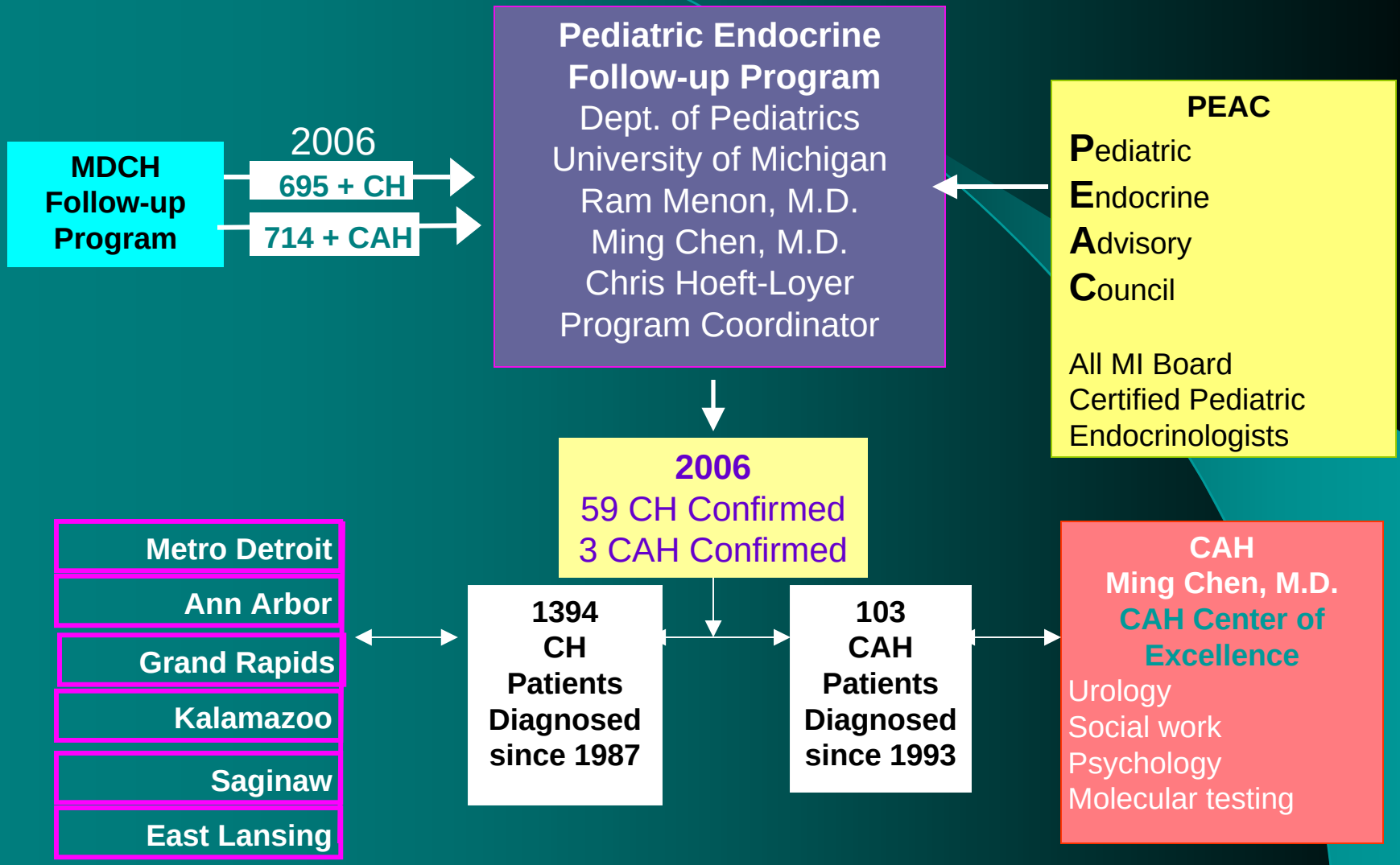
- Organic Acids
- Amino Acids
- Total and Free Carnitine
- Biotinidase Assay

Coordinated Care Plan	**Genetic Counseling**	**WIC/CSHCS Programs**
Emergency Care Plan	**Nutrition Services**	**Formula Distribution**
Social Work Services	**Nurse Practitioner**	**Psychological Testing**
	Home Monitoring	

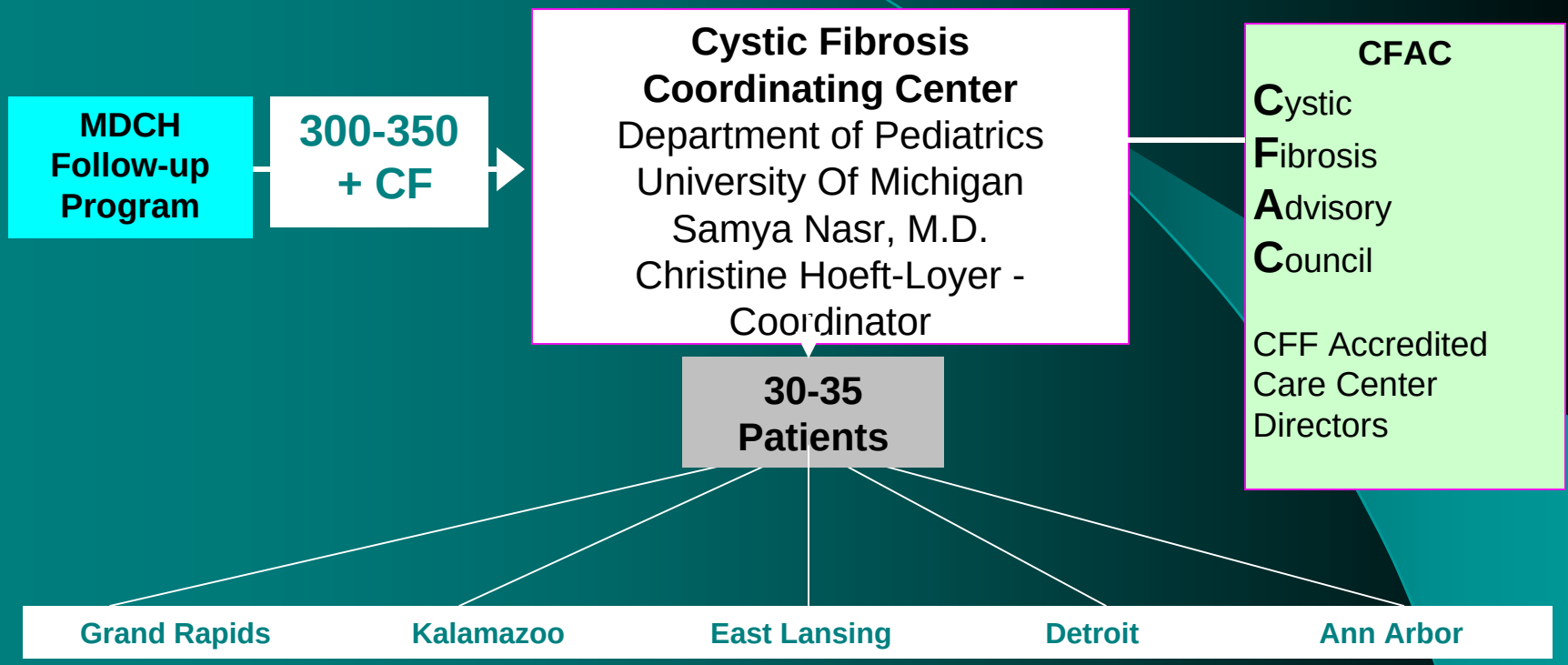
HEMOGLOBINOPATHIES FOLLOW-UP



ENDOCRINE FOLLOW-UP



CYSTIC FIBROSIS FOLLOW-UP



NBS PANEL EXPANSION

SEVERE COMBINED IMUNODEFICIENCY DISORDER (SCID)

Incidence: >10 genetic forms; 1/50,000 births; DiGeorge Syndrome; 1/4,000

Screening Method: T cell receptor excision circles (TRECS); IL7 **-** Wisconsin

Treatment: Cord Blood or bone marrow transplant

Gene therapy

NBS PANEL EXPANSION
LYSOSOMAL STORAGE DISORDERS

Incidence: 40 –50 disorders; 1/7,000 births
Screening Method: TMS-multiple enzyme assays

Treatment: Cord blood or bone marrow transplants (11):

- Globoid-cell leukodystrophy-Krabbe Disease – New York
- Cerebral X-linked adenoleukodystrophy
- Metachromatic leukodystrophy
- Hurler syndrome (MPS I)
- Maroteaux-Lamy syndrome (MPS VI)
- Sly syndrome (MPS VII)
- Gaucher disease
- Fucosidosis
- Alpha-Mannosidosis
- Aspartylglycosaminuria
- I-cell disease (Mucolipidosis II)

NBS PANEL EXPANSION

LYSOSOMAL STORAGE DISORDERS cont.

Treatment: Enzyme replacement therapy (6):

- Gaucher disease
- Fabry disease
- Hurler syndrome (MPS I)
- Hunter syndrome (MPS II)
- Maroteaux-Lamy syndrome (MPS VI)
- Pompe disease