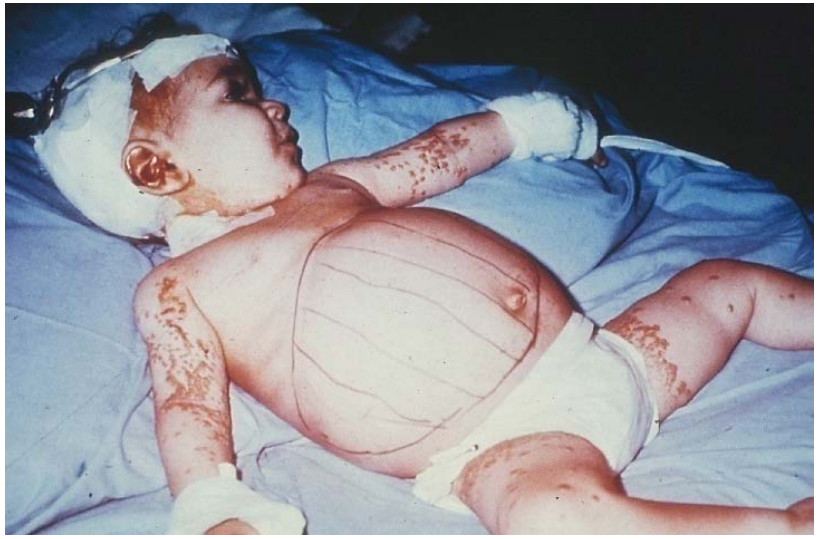
UConn / Connecticut Children's Medical Center Glycogen Storage Disease Program: Lessons for CT from a Rare Disease

David A. Weinstein, M.D, M.M.Sc.
Professor, University of Connecticut
Director, Glycogen Storage Disease Program

Background

Prior to 1971, glycogen storage disease type Ia was almost universally fatal marked by extreme failure to thrive, life-threatening hypoglycemia, and acidosis.



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Background

In 1982, cornstarch was introduced as a slow release glucose source

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Glycogen Storage Disease in 1998

- Most children in the United States and Canada were getting liver transplants or dying
- No clinical research was occurring
- 16 years without a major advancement

There was little hope for families

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Many Children Suffer Due to Lack of Knowledge About GSD

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Impact of Treatment

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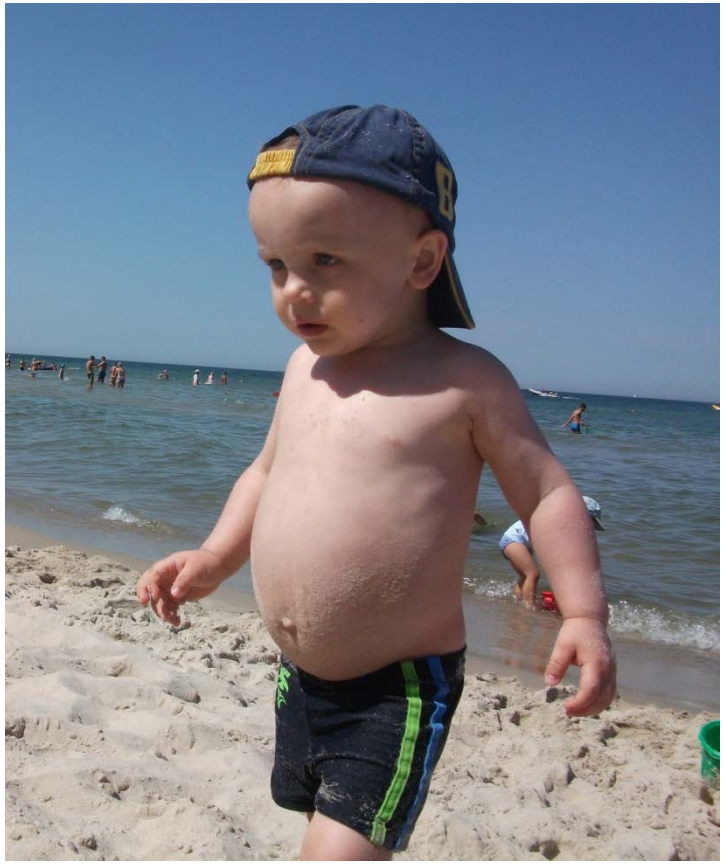
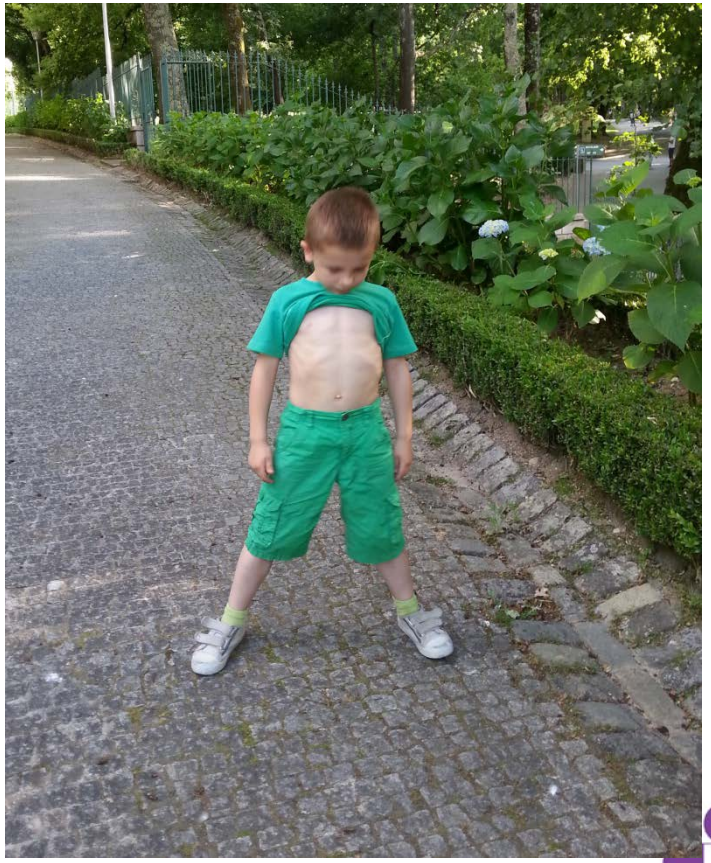
Impact of Treatment





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Impact of Treatment

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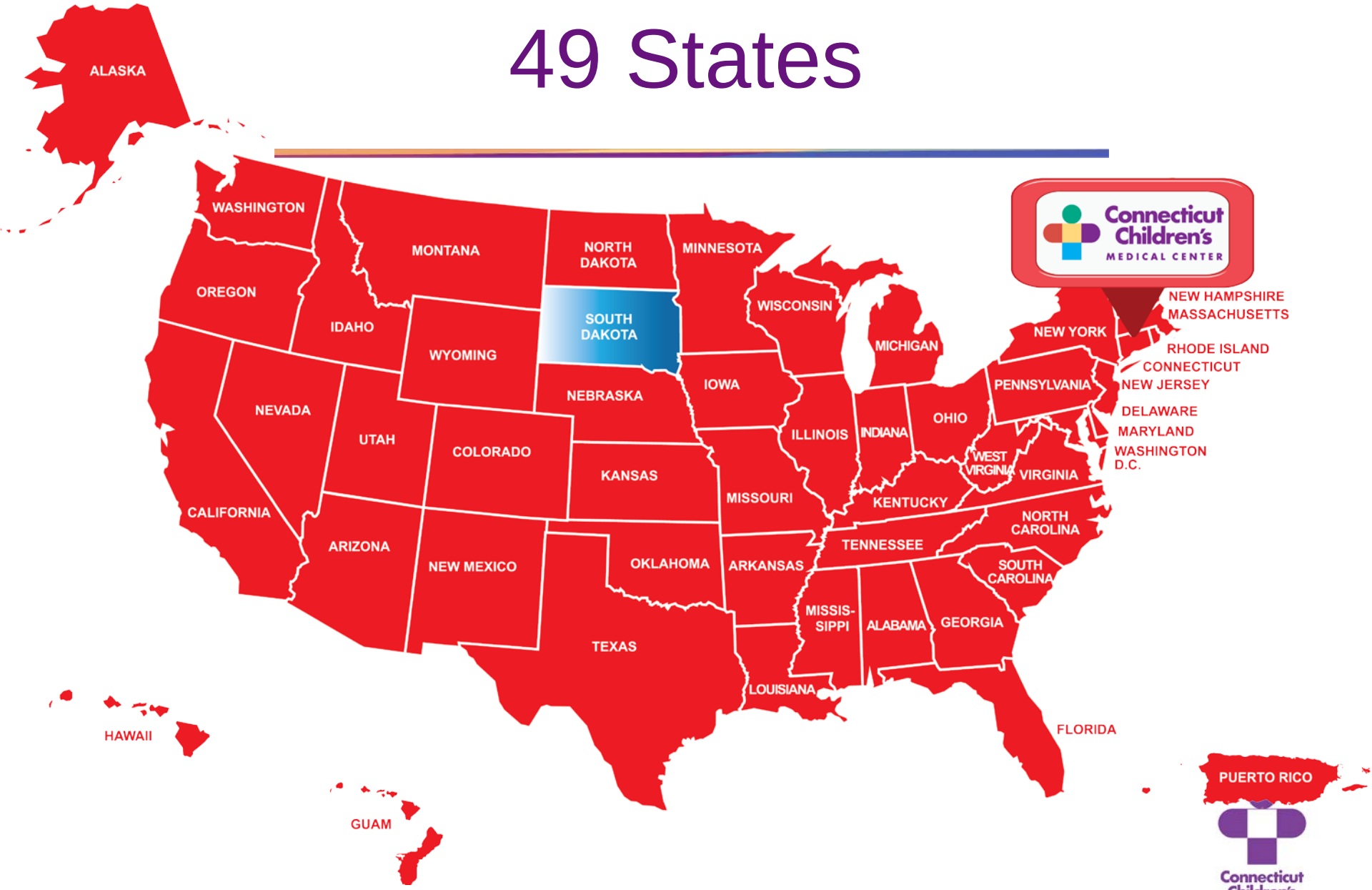
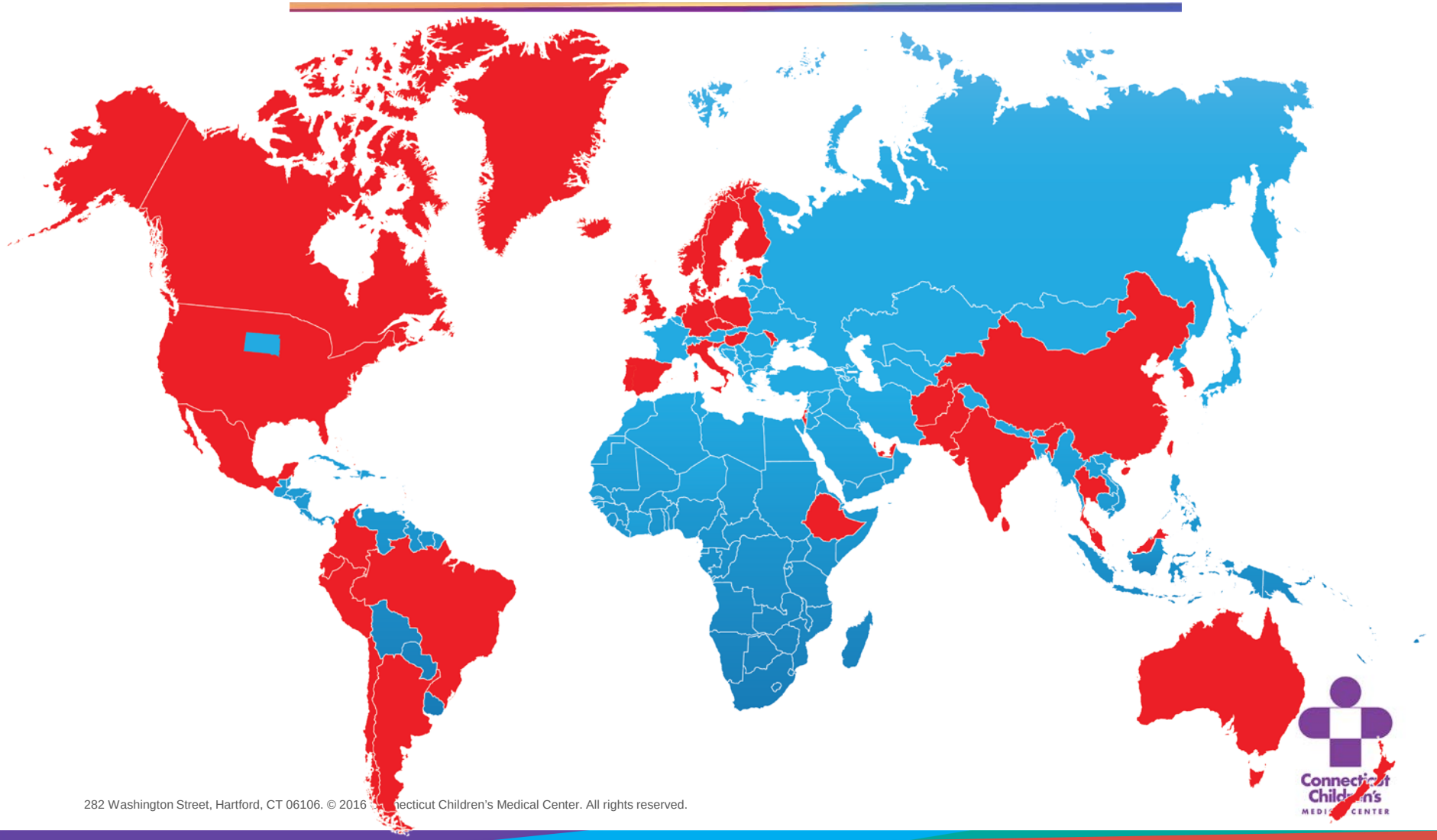
49 States



48 Countries



History

- Program previously located at Boston Children's Hospital (until 2005) and University of Florida (2005 – 2017).

- Program relocated to Connecticut in January 2017

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Rationale for Moving to CT

- GSD families built us clinical and research units at CCMC and UConn
- Joint collaboration between UConn Health and Connecticut Children's
- This would not have occurred if programs were not connected

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UConn GSD Gene Therapy Unit

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CCMC GSD Unit

- Located on the 8th floor of the hospital
- 3 bed clinical and research unit created
- GSD program office
- Clinical laboratory

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Impact on CT

- Over 200 families have traveled to CT for care and education in the first 10 months
- Over 1000 days of hotels and housing have been required
- 12 families have moved to CT and purchased housed in CT due to the program and this is estimated to increase to over 20 by 2018
- Students coming CT colleges due to the program

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Investment in Rare Diseases is Good for the State of Connecticut

- Patients and families will travel for rare disease specialists
- Investment by the state in rare diseases financially benefits the state
- Support for the medical institutions is critical

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