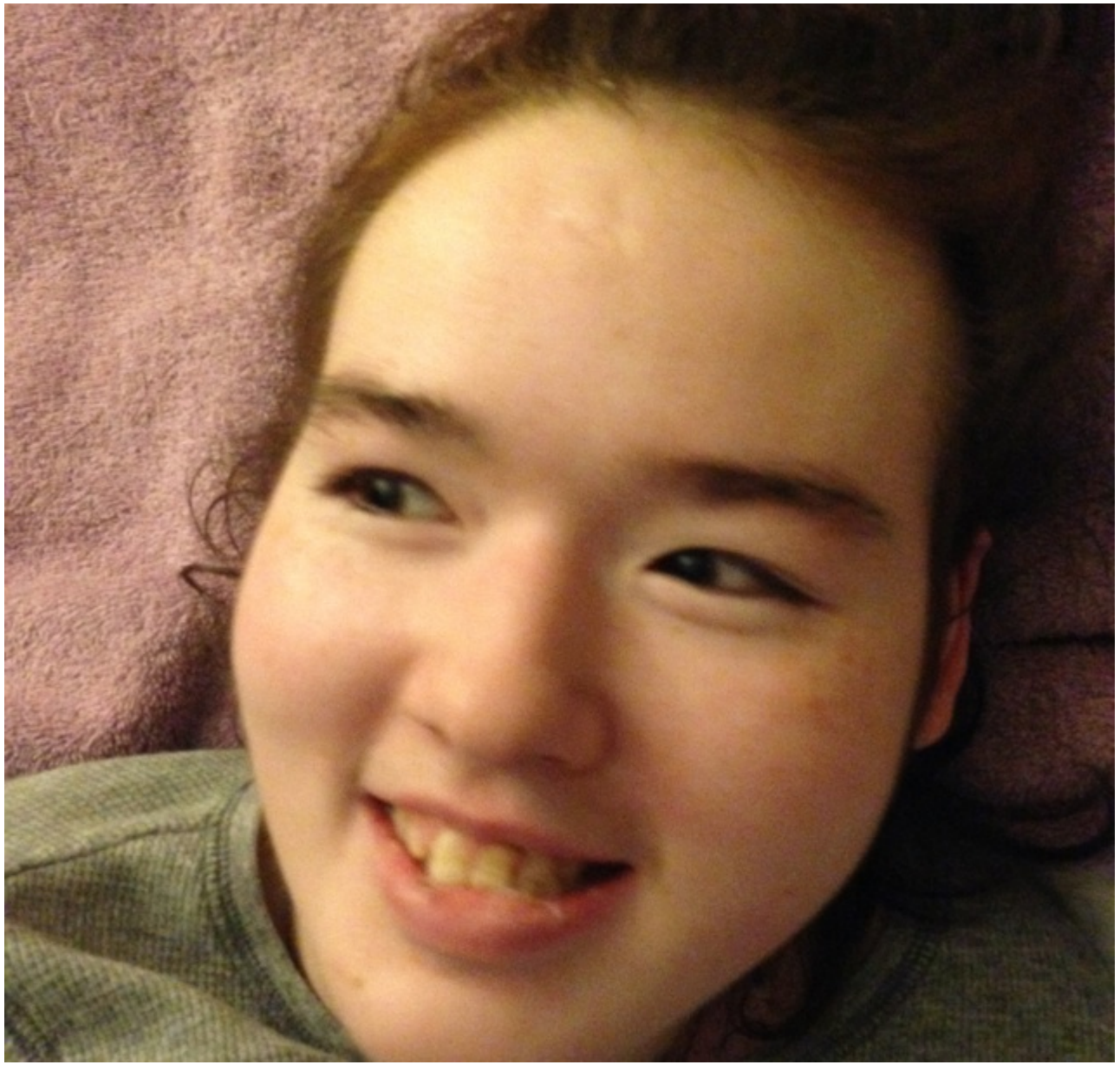
Kelly

Undiagnosed

Syndrome Without a Name



Connecticut Rare Action Network Community

- NORD estimate: >300,000 Connecticut residents have a rare disorder
- ~70 CT Rare Disease patient organizations, chapters, support groups
- >50 Connecticut companies engaged in work on rare disorders
- Yale, UConn, Quinnipiac, JAX Labs engaged in Rare Disease research
- Patient organizations fund state research and bring programs to CT

CT-RAN

- Network of rare disease patients and their families/caretakers, patient advocates, researchers in academic centers and industry, healthcare providers and lawmakers acting on state and federal issues that can impact the lives of those affected by a rare disease
- Increase awareness of elected officials of the emotional, physical, and financial burden that managing a rare disease places on the patient, the patient's family/caregivers, and the community
- Work with legislators to address issues of access and coverage for essential treatments, therapies, and services for rare disease patients
- Work to create a permanent Connecticut Rare Disease Advisory Council/Committee

Undiagnosed Patients:

medical outcasts in a modern health system

- *"Undiagnosed"* is a term used to describe a patient believed to have a rare genetic disorder that current testing has failed to identify.
- Diagnosis of a rare disease may take longer than 5 years…and some patients may never receive a diagnosis
- In the UK, it is estimated that ~6000 children are born every year with a genetic condition that physicians are unable to diagnose
- NIH experts estimate that 30-40% of the children with special needs in the USA lack a specific diagnosis
- Children with undiagnosed disorders often have complex medical conditions that include global developmental delays, seizures, neurologic impairments, feeding or breathing difficulties, physical disabilities
- Undiagnosed patients are often misdiagnosed and receive costly inappropriate care or medical treatments

Resources for Undiagnosed

- **NORD** web page for Undiagnosed
- **NIH**: Genetics & Rare Diseases Information Center (GARD), Office of Rare Disease Research (ORDR); Undiagnosed Disease Network (UDN); and Rare Diseases Clinical Research Network (RDCRN)
- **Genetic clinics/counseling**: University of Kansas list of genetic centers (kumc.edu): National Society of Genetic Counselors(nsgc.org), GeneTests (genetests.org), American College of Medical Genetics
- **Online genetic tools/programs**: Genome Connect, Rare Connect, MyGene2, and social media sites
- **Patient organizations**: Eurodis, Genetic Alliance, Global Genes, Syndromes Without a Name (SWAN), Rare & Undiagnosed Network (RUN)

Undiagnosed Programs and Studies

- University of Alabama Undiagnosed Diseases Program
- Children's National Health System (*Washington DC*) Undiagnosed Disease Study
- Columbia University (NYC) Discovery Program for Undiagnosed Diseases
- Undiagnosed & Rare Diseases Program, Children's Hospital of Wisconsin
- Scripps (CA) Idiopathic Diseases of Man (IDIOM) Study
- Pediatric Patients with Metabolic /Genetic Disorders Study (NIH)
- ???YALE

RDCRN

- **RDCRN** (NIH Office of Rare Diseases Research initiative) comprised of 21 rare disease research groups/consortia and a coordinating center
- **RDCRN** is a collaborative network of researchers and patient organizations working together to improve available information, treatment options, and general awareness of rare diseases
- **UDN**: is designed to accelerate discovery and innovation in the way we diagnose and treat patients with previously undiagnosed diseases.
- 2012: the successful NIH Undiagnosed Disease Program expanded and became a network of 7 clinical sites (Harvard, NIH-Bethesda, Duke, Vanderbilt, Baylor, UCLA, Stanford)
- **NIH** site in Bethesda continues to see ~150 patients/year and each of the other sites see ~50 patients/year