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
UndiagnosedPatients:
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 (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