



# LEVERAGING A RARE DISEASE CENTER OF EXCELLENCE

*This toolkit is a follow up resource based on the Global Genes Webinar **Leveraging a Rare Disease Center of Excellence** that was originally webcast June 1, 2015.*

*Our panelists included Kari Luther Rosbeck, President & CEO of the Tuberos Sclerosis Alliance; Mohamad Mikati, Chief of the Division of Pediatric Neurology at Duke University; Kelly Ranallo, Founder and President, Turner Syndrome Global Alliance; and Stephanie Okey, Head of North America, Rare Diseases, and senior vice president and General Manager of the US Rare Diseases business of Genzyme. A replay can be viewed at <http://globalgenes.org/june2015webinar/>.*

# LEVERAGING A RARE DISEASE CENTER OF EXCELLENCE



## Introduction

As many rare disease patients continue on their journey to become empowered advocates, some may want to consider starting a rare disease center of excellence. This toolkit covers the benefits the centers provide patients, steps that can be taken to establish a center, and what obstacles and challenges may need to be overcome to succeed.

Centers of excellence are valued by rare disease patients because of the complexity of their conditions and the need many of these patients face to seek treatment from a variety of different specialists. Centers of excellence provide an opportunity for coordinated care among a group of specialists, allow for easier access to these specialists, and connect patients and their families to others with the same disease.

Though some people may feel that establishing a center of excellence might present an insurmountable financial hurdle, the reality is that by engaging with a medical professional who can champion the idea within an institution, making a business case for establishing the center, and serving as a conduit to patients and other centers of excellence, it is possible to meet with success without facing any unusual financial burdens.

### Complex Diseases

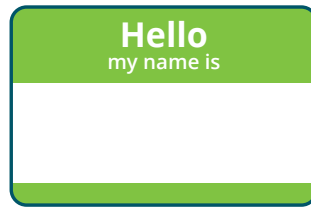
Rare diseases tend to be complex diseases that require a variety of specialists to provide appropriate care for a patient. Consider tuberous sclerosis complex (TSC), a rare genetic disorder that causes non-malignant tumors to form in different organs, including the brain, eyes, heart, kidney, skin and lungs. People with the disease can suffer seizures, developmental delays, intellectual disabilities, and autism.

“Individuals with TSC require a team of physicians, such as neurologists, cardiologists, nephrologists, dermatologists, pulmonologists—all the ologists—occupational, physical, and speech therapists; and of course, very strong partnerships with educators,” says Kari Rosbeck, president and CEO of the Tuberous Sclerosis Alliance.

There are many benefits to coordinated care and having specialists work together, especially in the case of rare diseases, where a complex of specialists are often needed to properly treat a patient. The coordination of care allows doctors to build a depth of understanding of a specific rare disease, its treatment, and manifestations. It also allows these specialists to more easily share information about patients, treat them more holistically, and benefit from each other’s understanding.

For patients, the benefits extend beyond the medical care itself. It is often difficult for patients to get access to the many specialists they need. Coordinating appointments and travelling long distances to see doctors can be a significant challenge and require long waits. One of the benefits of a center of excellence is having the necessary specialists available and usually being able to see the various doctors a patient needs to see in the same day at the same location or over consecutive days. Because of the way these clinics generally schedule patients around specific clinic days, another benefit they provide is bringing patients together and fostering connections between patients and their families. This brings together people from the same geographic region with the same disease and may put children in touch with other children with the same diseases for the first time. It also allows parents of children with a rare disease to connect with other parents and share experiences, information, and advice.

# SECTION 1: A ROSE BY ANY OTHER NAME



This toolkit uses the term “center of excellence,” but it should be noted that the term does not have a clear definition. Many so-called centers of excellence do not use the term to describe themselves, and so it is used in this toolkit to broadly describe the concept. In fact, none of the centers of excellence referred to in this toolkit use the term.

Some patient advocacy organizations, such as the Tuberous Sclerosis Alliance, provide designations through an application and review process to help patients understand the resources available at a clinic and provide standards to qualify for specific designations as a center for the treatment of a specific rare disease.

Despite the nebulous nature of the term, centers of excellence programs do share common elements. There is no minimum standard for a center of excellence, but below is a list of some common elements centers of excellence may include. A center of excellence may have some or all of these elements. Some rare disease patient advocacy organizations will look for certain elements they consider essential before considering a clinic a center of excellence.

## Core Elements

The UK Plan for Rare Diseases, which recognizes the need for centers of excellence,

outlined key characteristics that centers should have, according to Rare Diseases UK. These include:

- Coordinated care;
- Adequate caseload for expertise;
- Not dependent on a single clinician;
- Arrangement for transition from children’s to adults’ services;
- Engaged with people with rare conditions; and
- Research active.

## Setting Standards

The Tuberous Sclerosis Alliance has created a set of standards for clinics wishing to be considered Tuberous Sclerosis Complex clinics. This includes standards for clinical care, education, partnerships, and research. A complete description can be found on the Tuberous Sclerosis Alliance website’s clinic guidelines (<http://www.tsalliance.org/documents/TSC%20Clinic%20Guidelines%20October%202014.pdf>), but some of the key points are highlighted here.

The organization’s guidelines call for these clinics to have board certified healthcare providers that provide multidisciplinary care as a team for children, adolescents and/or adults. The director of the clinic should be a medical doctor with medical board certification in their country of clinical practice in a specialty related to one or more aspect of tuberous sclerosis complex, such as cardiology, dermatology, nephrology, neurology, or other area. While it requires a TSC Clinic to provide care in at least three core specialties, a TSC Clinic seeking to meet the organization’s higher Gold Standard must provide care in six or more specialties.

Beyond the medical care provided by these clinics, the organization expects clinics to play a critical role in educating patients, their families, educators, healthcare providers,

## SECTION 1: A ROSE BY ANY OTHER NAME

and the public about the disease and to work closely with the patient advocacy group to push out information and promote awareness of the disease.

Clinics are also expected to form partnerships with their local TSC group. This would include engaging volunteers from the organization to provide resource information and support for individuals with TSC and their families, and TSC Clinic staff will refer the individuals and families to the community alliance.

Regarding research, the organization expects that one or more the clinic's healthcare providers should either conduct investigator-initiated clinical trials in TSC, participate in a TS Alliance or industry sponsored TSC clinical trial, or conduct basic science research in TSC.



## SECTION 2: CREATING A CENTER OF EXCELLENCE

Centers of excellence are sometimes started by the work of a patient advocate or advocacy group and sometimes driven by the interests of a clinician. For a center to move from vision to reality though, it generally requires a partnership of both.

For patient advocate seeking to create a center of excellence, some key steps to success include:

- Find the right hospital or organization with which to partner;
- Find an internal champion;
- Make a business case to the provider;
- Understand the patient population in the area it will serve;
- Be a credible partner; and
- Be determined, but patient.

Consider the case of the Kansas City Great HeightTS Clinic at Children's Mercy Hospital in Kansas City, Missouri, which focuses on treating patients with Turner Syndrome. Turner Syndrome is caused by a partial or complete absence of one of the X chromosomes in girls and women. There is great variance in how the disease manifests itself. Among the long list of possible effects are delayed puberty, short stature, heart defects, infertility, scoliosis, learning difficulties, and improperly formed kidneys.

Kelly Ranallo, Co-Founder and President of the Turner Syndrome Global Alliance, was instrumental in the establishment of the clinic, a reflection that determination can be more important than money when it comes to creating a center of excellence.

Kelly's efforts began after her daughter, then 8, was diagnosed with Turner Syndrome and she found it challenging to access and coordinate the care her daughter needed. She identified Mercy Hospital in part because its ability to accept all payer sources, which meant that the broadest number of families would be able to access it. In an effort to gain influence with the hospital, she joined its family advisory board, which was established to provide patient feedback on how to provide better care.

At the same time, Kelly began looking for an internal champion at the hospital who could push for the establishment of the clinic from within. She found her champion in an endocrinologist who had an interest in Turner Syndrome. She also put together a business plan and started discussing it with the hospital to show that the clinic could work within the hospital's existing infrastructure and plan. She made a strong business case for the hospital. The clinic would not cost anything more to run, but it would provide economic benefits by decreasing the no-show rate of patients, and increasing the hospital's exposure by attracting families from six surrounding states that would drive to the clinic for care.

*"You have to be a credible partner, knowledgeable, and understand the mission and needs of the hospital," says Kelly Ranallo. "It's a marathon. It took me about four years from the time my daughter was diagnosed to the first clinic."*

## SECTION 2: CREATING A CENTER OF EXCELLENCE

### Don't Forget to Ask

Once a doctor who can serve as an internal champion is identified, Stephanie Okey, Head of North America, Rare Diseases, and senior vice president and General Manager of the US Rare Diseases business of Genzyme, suggests that the patient advocate sit down with them and write a list of all the specialties that a patient with the rare disease on which they are focused might possibly need to see.

Then, they should go to each of those departments and talk to the doctors one-on-one or have grand rounds. She says a patient and a healthcare provider should work together and put a face and name behind the condition.

“We’ve found it’s not just enough to go and talk about the disease in a grand rounds or a lecture-type setting, but end it by saying, ‘Who of you will help us? Who of you will work with us to provide comprehensive care?’ Just asking is so important,” says Stephanie.



## SECTION 3: FINANCING A CENTER OF EXCELLENCE



Though raising money to fund a center of excellence may seem like an easy way to ensure success, Kelly's approach didn't require her organization or the hospital to raise or invest capital to create and run the center. The benefit of this approach is that it builds into the center a business model that is sustainable. Other centers, she notes, that were dependent on raising money to establish themselves have proven less sustainable and continue to be reliant on raising money to continue their operations.

Mohamad Mikati, chief of the Division of Pediatric Neurology at Duke University, was successful at establishing an Alternating Hemiplegia of Childhood (AHC) center of excellence at Duke Children's Hospital & Health Center. AHC is a rare genetic neurodevelopmental disease that, among other things, can cause epilepsy, cerebral palsy-like movement problems, learning disabilities, behavioral issues, and episodes of temporary paralysis.

Mohamad says his effort to start the clinic was an acknowledgement of the need for a multidisciplinary team to treat patients. He was able to bring together colleagues with different expertise without access to extra funding.

*"To put together a multidisciplinary team is costly," Mohamad says. Initially, Mohamad brought together a variety of specialists to discuss AHC cases. Eventually, he says everyone was convinced that to address these patients a multidisciplinary team was needed and everyone came together without having*

*extra funding initially. "When people realize how important things are and that they can make a difference, you can go a long way with that."*

### Partnership: The Essential Ingredient

At the heart of successful centers of excellence often lies a partnership between clinicians and patient groups. In the case of Duke's Alternating Hemiplegia of Childhood center of excellence, the advocacy group Cure AHC played a critical role in its establishment and ongoing operations. "Cure AHC is very important for us, and has been from the beginning and will continue to be," says Mohamad.

The group did provide some financial assistance early on, but it also played a critical role in expressing the needs of patients to ensure that what the clinic did was meaningful. For instance, one thing the organization expressed was the need for physical and occupational therapy evaluations for patients. The group also plays an important role in making AHC patients aware of the clinic, serving as a liaison between the clinic and patients, and helping patients far away from its location in Durham, North Carolina with the logistics of getting there. All of that, says Mohamad, was critical in the initiation and success of the clinic.

*"They are the beginning and the end," Mohamad says of patient groups. "They are the ones who inspire us to start these clinics. They are the receivers of the care, the monitors of the care, and the guidance they provide is very essential."*

Patient groups, such as the Tuberous Sclerosis Alliance, also play an important role in connecting centers of excellence together, facilitating collaborations on research and fostering an exchange of ideas on best practices.

## ADDITIONAL RESOURCES



**Centres of Excellence for Rare Diseases, Rare Disease UK,**  
<http://www.raredisease.org.uk/documents/Website%20Documents%20centres-of-excellence-10-a4.pdf>

The Global Genes Webinar **Leveraging a Rare Disease Center of Excellence** that was originally webcast June 1, 2015 can be viewed at <http://globalgenes.org/june2015webinar/>.

The Tuberous Sclerosis Alliance and **Tuberous Sclerosis Complex Clinics Scope of Relationship Policy** can be found online at [http://tsalliance.org/documents/Scope%20of%20Relationship%20Policy%20\(Versions%201.5%20Aug%202013%20approved%2010.4.2013\).pdf](http://tsalliance.org/documents/Scope%20of%20Relationship%20Policy%20(Versions%201.5%20Aug%202013%20approved%2010.4.2013).pdf).

**The Tuberous Sclerosis Alliance TSC Clinic Network** TSC Clinic Guidelines can be found online at <http://tsalliance.org/documents/TSC%20Clinic%20Guidelines%20October%202014.pdf>.



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of This Toolkit's Contributors**

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