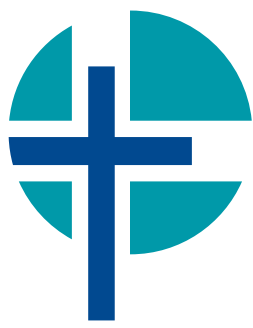


February
2021



SAINT PETER'S HEALTHCARE SYSTEM

Safely treating you better...for life.

Saint Peter's Assists Those with Genetic Rare Diseases

February 28th marks Rare Disease Day this year. The last day of February is annually designated to raise awareness about rare diseases and to educate others on how these diseases impact a patient's life. Improving the general public's knowledge about rare diseases can propel researchers and decision makers to better address the needs of those living with a rare disease. An estimated 30 million Americans (10% of the country's population) are affected by approximately 7,000 different rare diseases, according to the National Institute of Health's Genetic and Rare Diseases Information Center.

At least 80% of rare diseases are known to have a genetic cause, therefore, physicians may recommend genetic counseling or a genetic evaluation to their patients. As a state-designated regional center, the Saint Peter's Department of Medical Genetics and Genomic Medicine provides inpatient and outpatient services to patients of all ages with a suspected rare disease.



"Genetic disorders are often difficult to precisely diagnose not only because they are rare (affecting fewer than 1 in 1,600 people in the United States) or ultrarare (affecting fewer than 1 in 50,000 people) and numerous, but because their natural history is often poorly defined and their spectrum of presentation is broad. Complicating the matter further is the fact that confirmation of a clinical diagnosis is dependent on selecting the correct genetic test to order and properly interpreting the results in the context of the entirety of the patient's history and all their presenting features," said Debra-Lynn Day-Salvatore, MD, PhD, FAAP, FACMG, a renown geneticist and chair of the Department of Medical Genetics and Genomic Medicine at Saint Peter's.

Rare diseases present a diagnostic challenge not only for the medical practitioner but for the impacted individual. Many families spend months or years seeing a variety of specialists before they are appropriately referred to a medical geneticist. There are substantial burdens to not recognizing the presence or possibility of a rare disease. These include the physical burden for the patient from unaddressed symptoms, the emotional burden for the family of living with the unknown, the burden on the medical care system from unnecessary or redundant office visits and studies, and finally the financial burden from not initiating targeted management or treatment.

Many of the patients who come to see Dr. Day-Salvatore have been on a diagnostic odyssey, seeing multiple subspecialists to no avail. The genetics team reviews the clinical summaries of all previous providers before they see a patient. This review allows the specialists to see the "big picture." However, it is so very often something subtle, unique or rare that helps in making the diagnosis. Medical geneticists, such as Dr. Day-Salvatore, very often arrive at rare diagnoses by recognizing the importance of either a physical or laboratory finding that had previously been overlooked by others.

"Geneticists are especially equipped to diagnose because they are able to marry the medical with the technology and the psychosocial aspects, and carry the family through the diagnostic odyssey – from the initial questions to finally having some resolution," said Donna Chavez, MS, CGC, intake coordinator at Saint Peter's. "Families can be in angst for months or years worrying about what's wrong with their loved one. To finally have a team that's invested and educated and able to move the patient forward by ordering the appropriate studies to get them an answer can be quite gratifying."



**Debra-Lynn Day-Salvatore,
MD, PhD, FAAP, FACMG**

This year, Rare Disease Day will be celebrated virtually, which mimics the transitions made in the healthcare world this past year due to the ongoing COVID-19 pandemic. The Department of Medical Genetics and Genomic Medicine was not alone in making the transition to telehealth, however the staff was very grateful to already have the infrastructure in place having been the first department at Saint Peter's to pilot and successfully launch telemedicine.

"What we found through the quick transition during the pandemic was that both patients and providers didn't feel any sort of drop in emotional connection," said Kristin M. Levandoski, MS, CGC, genetic counseling supervisor at Saint Peter's. "Your facial and nonverbal expressions are a huge part of counseling and wearing a mask can hide some of that. During video appointments, patients can see my smile and know that I'm genuinely present with them in the moment."

Building and maintaining an emotional connection with patients is a top priority for the 'Gene Team' at Saint Peter's. One of the most unique and fulfilling aspects for staff working in the department is being able to provide continuity of care to patients and their families. With the ability to offer genetic services across many subspecialties, including prenatal, pediatrics, and adult care, the department is able to truly offer care across the lifespan.

This unique structure has allowed Dr. Day-Salvatore and her team to care for patients starting from the time their mothers were pregnant with them, through childhood, adolescence and adulthood. It has even allowed for the team to care for the children of their patients if they are diagnosed with the same rare disease.

"We're really in this for our patients so their rare diseases feel a little less rare. We want our patients to have access to the same health care. They need to feel supported throughout their journeys," said Levandoski.

Saint Peter's Genetics Department strives to help patients with genetic rare diseases by providing a level of care and caring that surpasses families' and referring providers' expectations. Dr. Day-Salvatore and her team are passionate about their profession, which results in their patients' desire to return year after year, generation after generation.

Telemedicine services continue to be provided, when appropriate, in addition to in-person counseling and consultation. To learn more, please visit: saintpetershcs.com/genetics

For more information about Rare Disease Day, visit: rarediseaseday.org

For more information about rare diseases, visit: rarediseases.info.nih.gov/diseases

Saint Peter's Nationally Recognized with Leapfrog Hospital Safety Grade of 'A' for Fall 2020

Saint Peter's University Hospital received an 'A' for the Fall 2020 Leapfrog Hospital Safety Grade, demonstrating the hospital's commitment to high quality patient care. A national distinction, the Leapfrog Hospital Safety Grade looks at publicly available measures of safety such as the hospital's performance in preventing medical errors, injuries, accidents, infections and other issues related to patients in their care. Developed under the guidance of a national panel of experts, the Leapfrog Hospital Safety Grade uses up to 27 measures of publicly available hospital safety data to assign grades to more than 2,600 U.S. acute-care hospitals twice per year. The Hospital Safety Grade's methodology is peer-reviewed and fully transparent with results free and available to the public.

"Safety is always a number one priority at Saint Peter's but receiving this recognition amidst a pandemic is even more meaningful," said Leslie D. Hirsch, FACHE, president and CEO of Saint Peter's Healthcare System. "Receiving Leapfrog's highest safety grade reinforces patient confidence in the ability to receive quality medical care without compromise. At Saint Peter's, we are proud of our ongoing ability to safely deliver the highest quality, evidence-based clinical practices along with the compassionate care that is the hallmark of our Catholic mission."

The Hospital Safety Grade is compiled under the guidance of the nation's leading experts on patient safety and is administered by The Leapfrog Group, which for nearly 20 years has collected, analyzed, and published hospital data on safety, quality, and resource use. The Hospital Safety Grade is available free to the public with the goal of empowering people with the information they need to make better, informed healthcare decisions.

"Every single person at Saint Peter's plays an integral role to the success of the Leapfrog Hospital Safety Grade," said Sara Slate, BSN, RN-BC, CPHQ, performance improvement coordinator in the Quality and Safety Department at Saint Peter's. "Whether they are directly involved in the survey process or not, achieving an 'A' is not possible without the support of the entire healthcare system."

The Leapfrog Group's letter grade scoring system allows consumers

to quickly assess the safety of their local hospital and choose the safest hospital to seek care. To see Saint Peter's University Hospital's scores as they compare nationally and locally, and to find safety tips for patients and families, visit the Leapfrog Hospital Safety Grade website at www.hospitalafetygrade.org.

Exceptional Safety



for Exceptional Times

Community Calendar

For more information or to register for a program, call the contact number listed or visit saintpetershcs.com/Events. Most programs are **FREE** unless noted; please call for information.

Cancer Support

- Bone Marrow Transplant**
For patients who have had an allogeneic transplant
Call for information: 732.235.8522
- Breast Cancer**
Call for information: 732.235.7011
- Gynecologic Cancer**
In partnership with the Cancer Support Community Central New Jersey
Call for information: 732.235.6792
- Living with Cancer**
Call for information: 732.235.7557

Diabetes Education*

- For more information about the following classes, please call the phone numbers listed below.
- Diabetes Self-Management Education**
Call for information: 732.339.7630
 - Gestational Diabetes Self-Management Education**
Call for information: 732.339.7630
 - Type 1 Diabetes Support Group for Children and Their Families**
Call for information: 732.745.8600, x8610

Support Groups

- Bereavement**
Call Pastoral Care for information: 732.745.8565
- Craniofacial Teen Girls**
Call for information: 732.745.8600, x8113
- Spouse/Partner Caregiver**
Call for information: 732.745.8600, x8662

SHARE 3 rd Thursday of each month Next meeting: February 18 7 to 9 pm Irina Mariano-Brown, MSW, LCSW Email for Zoom link: imariano@saintpetersuh.com Call 732.745.8522 FREE	Weight Loss Seminars 3 rd Tuesday of each month Next meetings: February 16 English 12 to 1 pm Spanish 1 to 2 pm For information call 732.745.8571	MyGoal Autism Family 2 nd Sunday of each month Next meeting: February 14 3 pm <i>In-person (socially distanced seating)</i> Call 877.886.9462 or email info@mygoalautism.org for location details
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Parent Education

Parenting classes are currently being held online. Register at saintpetershcs.com/Services/Parent-Education. Once registered for a class, access information will be provided to you. For information about the following classes, please call Parent Education at 732.745.8579

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| Baby Care | Grandparents Class | New Mom Support Group |
| Birth Center Class
(for moms under midwifery care) | Hypnobirthing | Prenatal Yoga & Exercise Class |
| Breastfeeding Class | Marvelous Multiples | Prepared Childbirth/Lamaze Classes |
| Breastfeeding Support Group | New Daddy Class | Sibling Class |

"Lunch 'N Learn"
East Brunswick Public Library and Saint Peter's virtual session.

Arthritis

Friday, February 5 • 12 p.m.
Registration is requested for this free event.
For more information or to register, visit ebpl.org/calendar

Adolescents and Ways to Safely Socialize

Presented by Dr. Susan Brill
February 10 • 6:30 PM
Visit saintpetershcs.com/Events to register.

Need a doctor? To find a Saint Peter's physician, call 1.855.SP.MY.DOC (1.855.776.9362).
Online scheduling is also available with select Saint Peter's Healthcare System physicians via Zocdoc at BookMySPDoc.com
This calendar is for information only. Do not rely on it to make healthcare decisions on your own; instead, talk with your healthcare provider for advice and treatment. Do not ignore medical advice or delay seeking it because of something you read here. We do not suggest or endorse that you have any tests, use products, or use other information in this document without advice from your healthcare provider.



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