Medical Xpress

Down syndrome research gets a boost with new biobank initiative

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Nationwide Children's Hospital and research advocacy group DownSyndrome Achieves have joined forces to create the first biobank in the country dedicated to collecting and managing blood samples from people with Down syndrome.

The biobank will be housed in the Nationwide Children's Biopathology Center (BPC), which also houses the biobank for the Children's Oncology Group, The Cystic Fibrosis Therapeutics Development Network and the Nephrotic Syndrome Study Network, among others.

"This is a prime example of how we are using our expertise in biobanking to help researchers everywhere do clinical and translational research," says Daniel Coury, MD, chief of Developmental/Behavioral Pediatrics at Nationwide Children's.

The biobank was conceived and spearheaded by Lito Ramirez, founder of DownSyndrome Achieves and proud parent of a son with Down syndrome.

"Although Down syndrome affects approximately 250,000 Americans, it is one of the least funded research areas at the National Institutes of Health. It ranks in the bottom 20 percent of funding categories—much lower than cystic fibrosis, cancer and other genetic disorders," says Ramirez.

The biobank will begin by storing blood samples from Nationwide

Children's with plans to expand collection to other institutions and tissue types.

"Biobanks have been around since the 1990s," says Ramirez. "Nearly every research category except Down syndrome has had a centralized biobank. Now, we are able to support researchers by providing a national, centralized repository of samples donated by people with Down syndrome."

Certain conditions such as congenital heart disease, childhood leukemia and Alzheimer's disease are more common in patients with Down syndrome than in the general population. Other conditions, including solid mass tumors such as breast cancer and prostate cancer, high cholesterol and atherosclerosis are less common. Studying these conditions in the context of Down syndrome genomics will likely have implications for everyone.

At the Mass General Hospital Down Syndrome Program, translational and clinical researchers are investigating several conditions that accompany Down syndrome, including obstructive sleep apnea, thyroid disease and celiac disease.

"A biobank provides researchers with the potential to unlock some of the mysteries associated with Down syndrome," says Brian Skotko, MD, MPP, a medical geneticist at MassGeneral and director of the Down Syndrome Program there. "Currently, researchers do not have readily available and curated blood and tissue samples to advance our understandings of the conditions that co-occur with Down syndrome. I am excited that the Down syndrome community will have this national resource, which is long overdue."

Provided by Nationwide Children's Hospital