

BANKING FOR THE FUTURE

BY TARA SCHMIDLEN, MS, CGC

Patients and families affected by rare diseases are often eager to participate in research or contribute to science in some way, but may have limited options due to the rarity of these conditions. One way that patients and families can contribute to scientific research is by donating a blood/and or tissue sample to the NIGMS Human Genetic Cell Repository at the Coriell Institute for Medical Research in Camden, N.J. The NIGMS Repository seeks samples from people with inherited genetic diseases or chromosomal abnormalities for use in research. The NIGMS Repository is supported by the National Institute of General Medical Sciences, a branch of the National Institutes of Health (NIH). The Coriell Institute is an independent non-profit research organization dedicated to understanding human genetic diseases and providing the highest quality genetic resources. Donating a blood or tissue sample to a cell repository, like the NIGMS Repository at Coriell, directly supports and makes research into genetic diseases and chromosomal abnormalities possible.

So, what exactly is a cell repository and how are cell repositories important to the success of research?

A cell repository, or biobank, is a bank that stores cell lines made from human blood or tissue for use in research. A cell line is a population of cells that can continuously grow and divide. To make a cell line, living human cells are taken from a blood or tissue sample and placed in a container with a growth solution. This process is called cell culturing. The cells are allowed to grow and divide for a period of time. Eventually, groups of cultured cells are “harvested” and placed into small glass ampoules. The glass ampoules full of cells are then frozen in liquid nitrogen tanks at -316° F where

they are stored until requested by a scientist.

Cell repositories like the NIGMS Repository at Coriell have become a key money and time saving resource for scientists. It takes a lot of resources for scientists to find a group of people with a specific genetic disease or chromosomal abnormality and



CORIELL INSTITUTE
FOR MEDICAL RESEARCH

Coriell Institute for Medical Research, founded in 1953 and based in Camden, New Jersey, is an independent non-profit research center dedicated to the study of the human genome. Expert staff and pioneering programs in the fields of personalized medicine, cell biology, cytogenetics, genotyping, and biobanking drive our mission.

collect samples and medical information from each person each time a new research study is to be performed. The availability of a centralized source of existing, high quality cell lines becomes ever more critical for those scientists studying rare conditions which often have limited

funds for research.

Rather than expend precious time and resources collecting samples, qualified scientists from all over the world can access the cells and corresponding medical information banked at Coriell and use them for a variety of research purposes, including: discovery of new genes, studying how cells from individuals with genetic diseases or chromosomal abnormalities function, developing new ways to detect genetic diseases or chromosomal abnormalities, and the development and testing of potential treatments or cures. When an individual donates a sample to the NIGMS Repository, they are providing scientists all over the world



FREEZING THE FUTURE Cultured cells are harvested and are placed in glass ampoules (*left*) and then stored in liquid nitrogen tanks at -316°F (*right*).

with the very valuable opportunity to learn more about their specific disease and how their specific genetic make-up influences the symptoms and behaviors associated with their diagnosis.

The NIGMS Repository has been funded by the NIH since 1972 and is the world's largest public human genetic cell repository. The NIGMS Repository contains over 10,600 samples, representing over 500 different genetic diseases and chromosomal abnormalities. There

are many different types of cell repositories. Some focus only on a single disease of interest while others may focus on a group of diseases like cancers or muscular dystrophies. Some repositories are small, privately funded collections and their samples are only made available to a specific group of researchers. While both public and privately held repositories are valuable for research, one advantage of donat-

ing a sample to a public repository is that the samples are able to be accessed and utilized by multiple scientists from all over the world simultaneously, which can accelerate research progress and foster collaboration among groups of scientists. Samples donated to the NIGMS Repository

at Coriell have been used in over 5,500 scientific publications by scientists from over 50 different countries.

In addition to collecting blood and tissue samples, the NIGMS Repository also

collects information about the medical and family history of the sample donor. While genetic test results are not required for a sample to be added to the repository, the more information that accompanies a sample, the more useful that sample is to researchers. We take the privacy and confidentiality of each donor very seriously. To protect the privacy of donors, samples are coded with a number and all names,

dates of birth or other potentially identifying information are removed from both the sample and any accompanying medical records. Only limited demographic information is provided to researchers who access the cell lines and includes the age of the donor at the time of sample collection, gender, diagnosis, race/ ethnicity and country of origin.

One exciting approach to learning more about genetic diseases and chromosomal abnormalities that capitalizes on cell lines banked in the NIGMS Repository is the development and use of man-made stem cells, called induced pluripotent stem (iPS) cells. iPS cells are man-made embryonic-like stem cells that are made by taking mature human cells (e.g. skin cells) and reprogramming the cells back to an embryonic, undifferentiated or "blank-slate" state.

These "blank-slate" iPS cells can be directed to develop into any cell type so that scientists can test how well a drug or therapy may work on those cells. Using iPS cells allows scientists more opportunity and easier access to study difficult to obtain target tissues like muscle or nerve tissue. Scientists can make unlimited

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The screenshot shows the Coriell Institute website interface. At the top, there is a search bar with the text 'duplication chromosome 15' and a 'GO' button. Below the search bar, there are navigation links: Home, News, Browse Catalog, Collections, Services, and Contact Us. The main content area displays 'Search Results For: duplication chromosome 15'. It includes filter options for 'Catalog Items (58)' and 'Catalog Documents (14)'. There are sections for 'Filter Results by Collections' (with 'NIGMS Human Genetic Cell Repository (58)' selected), 'Filter By Products Available' (with 'All' selected), and 'Display Optional Fields (Check to display)' (with 'Karyotype', 'Family Number', 'Relationship to Proband', 'Ethnicity', 'Reported Mutations', 'Affected Status', and 'Publications Cited' all unchecked). Below the filters is a table of results. The table has columns for Catalog ID, Description, Cell Type, Cell Line, DNA, Panel, Genotypes, Sex, Age, Race, and PDL. The table shows 12 results, with the first row being GM04347 (INVERTED CHROMOSOME) and the last row being GM23126 (DUPLICATED CHROMOSOME). The table is paginated with 'Page(s) 1 2 3 4 5'.

Catalog ID	Description	Cell Type	Cell Line	DNA	Panel	Genotypes	Sex	Age	Race	PDL
GM04347	INVERTED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Female	38 YR	Caucasian	
GM22397	DUPLICATED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Male	16 YR	Caucasian	
GM22394	DUPLICATED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Male	6 YR	Caucasian	
GM21715	DUPLICATED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Male	7 YR	Black	
GM23235	DUPLICATED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Male	4 YR		
GM22894	DUPLICATED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Female	11 YR	Caucasian	
GM22571	DUPLICATED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Male	12 YR	Caucasian	
GM23081	DUPLICATED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Male	5 YR		
GM12134	PRADER-WILLI SYNDROME, PWS	B-Lymphocyte	Yes	NA12134	No	No	Male	15 YR	Caucasian	
GM22593	DUPLICATED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Female	3 YR	Caucasian	
GM23181	ISODICENTRIC CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Male	3 YR	Caucasian	
GM23126	DUPLICATED CHROMOSOME	B-Lymphocyte	Yes	Inquire	No	No	Female	6 YR	Caucasian	

SEARCH SUCCESS Scientists from all over the world can use the NIGMS Repository's online catalog to find samples from individuals with genetic diseases and chromosomal abnormalities.

quantities of "affected" iPS cells to study from a single skin biopsy. Having the ability to make unlimited quantities of iPS cells may be helpful for transplant-based therapies or to test the effects of several new candidate drugs for the treatment of a disease. In addition, iPS cells may offer scientists a better disease model since animal models do not always overlap human physiology.

Using iPS cells, a scientist can test the effect of a candidate drug directly on the target tissue/cell type (like human muscle or nerve cells) which could make recruiting enough physicians and research subjects to conduct clinical trials easier if the drug or therapy has been developed using the best possible model (by using iPS cells versus an imperfect animal model). iPS

cell technology may also help advance the development of promising new drugs or therapies. The better the quality of drug and therapy development testing, the more likely the drug or therapy is to win the approval of the FDA, support from insurance companies, and the interest of potential manufacturers and distributors.

Each sample that is donated to the NIGMS Repository is an important contribution to the acceleration, improvement and advancement of scientific research on genetic diseases and chromosomal abnormalities. The NIGMS Repository accepts sample donations from patients and families from all over the world and collects samples from affected individuals and both biological parents, whenever possible.

Sample donation requires the completion of an informed consent form, a submission form, and a clinical information summary form. Copies of genetic test results and physician summary letters are also requested, as the more clinical information that accompanies a sample, the more useful the sample is to researchers. Coriell covers the cost of shipment both ways and is able to reimburse sample donors up to \$40 USD for the cost of a blood draw. It is recommended that blood samples be collected during a scheduled doctor's visit and that tissue samples be collected during another planned surgical

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procedure in order to avoid incurring any sample collection fees. Coriell is not able to reimburse sample donors for any costs associated with having a tissue biopsy. Interested donors are not required to provide both a blood and a tissue sample, although both sample types are welcomed from donors when collection of both sample types is possible. The NIGMS Repository is not able to tell individual sample donors how their sample was used or who obtained materials made from their sample. Sample donors will not receive results of any testing performed on their sample or materials made from their sample. •

ABOUT THE AUTHOR:

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TAKE PART
Interested sample donors may contact Tara Schmidlen, MS, CGC, Genetic Counselor for the NIGMS Repository, at 1-856-757-4822 or tschmidl@coriell.org to see if they are eligible to participate and to learn more about this opportunity.