



Relationships, Newborn Screening in Chile, and the need for a Mass Spectrometer

FORWARD BY OZZY SAMAD

Your support for BBF's humanitarian response to the war in Ukraine has enabled us to make a meaningful impact on the lives of so many who are facing life threatening circumstances. An unanticipated positive outcome of our efforts has been the development of collaborative relationships impacting other areas of BBF's work.

One such partnership with neurologists at Boston Children's Hospital (BCH) and their colleagues in Chile, has brought forth the importance of Newborn Screening (NBS) and the lack thereof, even in relatively developed countries such as Chile. The goal behind the project brought forth first to BBF by Dr. Christina Brisco-Abath and Dr. Agnes Kielian from BCH, is to help acquire a Mass Spectrometer to be able to do such screenings in Chile, and thereafter, expand the project to developing countries in similar circumstances.

The article below explaining NBS was especially written for our readers by Dr. Harvey Levy who is a distinguished neurologist at Boston Children Hospital and

lectures at the Harvard Medical School, and Dr. Juan Francisco Cabello, a neurologist at the University of Valparaiso in Chile and Director of their Inborn Errors of Metabolism Laboratory. The aim of the authors is to point out the importance of newborn screening and the critical part the mass spectrometer can play in early identification and management of often life altering health issues.

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Relationships, Newborn Screening in Chile, and the need for a Mass Spectrometer

I first met Bryan when he was 34 years old. He was in an institution for the mentally retarded (as they existed in those days). He had no speech, and he was so severely hyperactive as to be almost unmanageable. His IQ was untestable but estimated to be less than 30. Bryan was brought to the clinic to find out if anything could be done to make it easier to manage him. When Bryan was tested in the clinic, he was found to have a very high level of phenylalanine, the amino acid in his blood that indicated he had the metabolic disease known as phenylketonuria (PKU). PKU is the first discovered and the most frequent biochemical cause of severe mental disability. Had Bryan's PKU been detected when he was a newborn, he would have been normal because he would have been placed on a special diet that prevents the brain damage in PKU. But he was born before newborn screening had been developed and by the time he was even 2 years old he had already developed the severe irreversible brain damage that was so evident as an adult.

screening was only for the one disease, PKU, and very soon Dr. Guthrie learned that there were many other metabolic diseases like PKU that caused brain damage or even, in these other diseases, heart damage or liver destruction or bone disease or other problems that could be prevented if babies were detected by a blood test just after birth before any signs of this damage appeared. However, newborn screening did not detect these other diseases because it only tested the blood for phenylalanine and these other diseases had had increases in other chemicals such as leucine or methionine or galactose. So, Dr. Guthrie modified his test so that it could also show an increase in leucine if the baby had maple syrup urine disease or an increase in methionine if the baby had homocystinuria. Now newborn screening could detect these other diseases as well as PKU.

The problem is that there are close to a hundred or more serious metabolic diseases that cause irreversible problems and need to be detected in the newborn

and Dr. Guthrie's modifications of his test could only test for two more. But in the late 1990's a very important development occurred. A technology known as tandem mass spectrometry was modified so that it could be used to test the blood from babies to detect with one test any one of 30

or 40 metabolic diseases. Within a few years a number of newborn screening laboratories were using tandem mass spectrometry and identifying babies for immediate treatment to prevent the problems

that otherwise would have occurred. By 2006 or 2007 every newborn screening program in the United States was using tandem mass spectrometry to greatly expand newborn screening and now it is also used in almost every western European newborn screening program as well as in Australia, New Zealand, Japan, and South Korea.

It is estimated that less than a third of newborns in the world have access to any newborn screening test.

The tragedy, however, is that in many other places throughout the world newborn screening is not performed and, consequentially, many babies are doomed to grow up to be like Bryan. It is estimated that less than a third of newborns in the world have access to any newborn screening test. In Latin America only 4 countries have programs with wide coverage (Cuba, Chile, Costa Rica and Uruguay) but many countries do not carry out any type of screening now more than 60 years after screening began in developed nations.

In times where global and equity in access to health seem to be concepts that generate high interest, it is entirely reasonable to generate efforts at different levels that allow improving access to these types of benefits, understanding that it is not enough to carry out only a physical examination of the newborn babies, but also to select the diseases that need to be incorporated in each area of the world and generate the conditions that would result in these babies receiving the treatments they require to grow into normal and healthy and productive children and adults. ■



Levy HL. Robert Guthrie and the Trials and Tribulations of Newborn Screening. Int J Neonatal Screen. 2021 Jan 19;7(1):5..

Newborn screening for PKU was developed by Dr. Robert Guthrie in 1961 and begun in Massachusetts in 1962. Within a few years it spread throughout the United States and into a few countries in Europe. But

A Brighter Future with Solar Power

BY KAITLYN NUBEL

It's official: Brother's Brother Foundation is powered by the sun.

After a 20-month long process to convert to a sustainable form energy, BBF "Flipped the Switch" in September and converted to solar energy. Connected Energy Group, a local Pittsburgh company, installed solar panels on the roof of BBF's warehouse that are expected to offset 92% of its energy needs and save the organization \$20,000 in fuel costs each year. BBF will also receive

other distinguished guests, including State Representative Aerion Abney of District 19.

Representative Abney said BBF's switch to solar is part of a "new era in Pittsburgh" in which old warehouses and steel mills are being repurposed.

The burning of fossil fuels creates toxic pollution, which has an impact on global health. In 30 years, the average rooftop solar panel system can reduce pollution by 100 tons of carbon dioxide! This means cleaner air for everyone on earth.

a rebate from the Pennsylvania Department of Environmental Protection for installing six electric vehicle chargers for public use.

The project came to fruition through a nearly \$179,000 grant from Green Mountain Energy, a renewable energy retailer that also funds nonprofits' sustainable efforts through its Sun Club program.

The ceremony to celebrate BBF's switch to solar was attended by representatives from Sun Club, BBF board members, and

"When I think about the legacy we're trying to leave for our next generation, it makes me feel confident as a father of two kids that the environment and the sustainability of our environment are going to be in a positive situation 10, 20 and 30 years down the line because of the decision that we're making here today," Abney said.

BBF President Ozzy Samad hopes the newly installed solar panels

and electric vehicle chargers are just the beginning of future sustainability efforts at BBF.

"This has only just begun. We hope to convert our truck to electric energy and install electric chargers in our second building down the street." Samad said. ■





Cover With Address Label

The Season of Giving is Here

The cooler air of late fall has set in and signs of the upcoming holidays surround us. The season of giving is here! In the Brother's Brother Foundation Development Office, we have been planning for our end of year fundraising campaigns since early August and we are excited to finally share them with you.

Brother's Brother Foundation will once again participate in Giving Tuesday. This day of giving began in 2012 and has since grown to an international movement. Last year an estimated 35 million adults participated in GivingTuesday 2021, and this year's participation is projected to surpass it. If you would like to join this generosity movement, make your donation online at www.brothersbrother.org/donate on Giving Tuesday, November 29th.

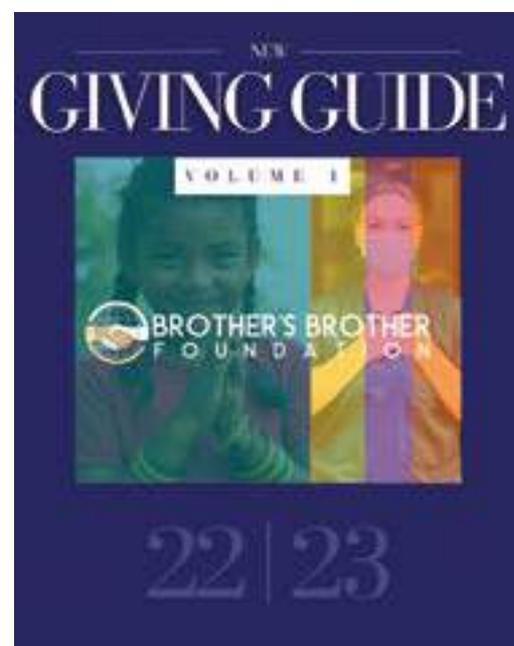
Be on the lookout in your email for our first ever Brother's Brother Foundation Giving Guide magazine. The new publication highlights our work in 2022 and provides a glimpse into what we have planned for 2023. We know you'll be inspired by the stories inside and hope the Giving Guide finds a home on your coffee table or magazine rack for the holidays.

Brother's Brother Foundation accepts gifts in honor and in memoriam as well. Give an especially meaningful gift this holiday by donating for someone else.

Give to Brother's Brother Foundation while you holiday shop! Amazon Smile is one of the easiest ways to support us. Select BBF as your charity and start from smile.amazon.com.

Thank you for your continued support of Brother's Brother Foundation!

We are sincerely grateful for each one of our donors – you make our work possible! ■



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BBF continues to be part of the Combined Federal Campaign. Many thanks to both new and renewing CFC donors.

Look for BBF on the Combined Federal Campaign website at <https://cfcgiving.opm.gov>. BBF's Combined Federal Campaign number is 12228. For donors who are not government employees, please remember that some employers match individual donations. Ask about your employer's matching gifts program.

This is a great way to give to BBF. Thank you!

