EXAMPLE 26 - 28, 2022

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Thank you Chan Zuckerberg Initiative

for recognizing the TANGO2 Research Foundation as a patient-led organization that is making a positive impact on the lives of patients and families affected by TANGO2- related disease



THIS CONFERENCE IS PARTIALLY FUNDED BY THE CHAN ZUCKERBERG INITIATIVE.

THANK YOU



FOR YOUR COMMITMENT AND SUPPORT OF OUR FAMILIES



THANK YOU



TOKIO MARINE HCC

FOR YOUR MISSION

"To support programs and institutions that enrich the quality of life with a particular focus on initiatives that help individuals, families and communities benefit from these services and opportunities. Our primary goal is giving back to communities where our customers, employees, and associates live and work."



INTRODUCTION

We are proud to welcome you to the 2nd TANGO2 Family Conference! We are happy to bring together the TANGO2 disease community for insights, fellowship and discussion.

Our mission is to improve the lives of those children and young adults affected by TANGO2-related disease by helping to fund, coordinate and guide scientific research that leads to a better understanding of how TANGO2 mutations affect them.

For many families and children affected by the rare disease, this conference will be one of the first opportunities to meet and engage with other affected families. Building these relationships is vital for combining efforts to advocate for research, treatment, and therapy for this rare disease. Thank you for joining us and we hope you will enjoy your experience and time with us.

- Conference Planning Committee

A MESSAGE FROM OUR FOUNDERS MIKE AND KASHA MORRIS

We'd like to welcome everyone to the 2022 TANGO2 Family Conference. Like many of you, as parents to a child struggling with TANGO2 disease, we wish we could somehow fast-forward time and have all the answers we seek today. Then, we pause and recognize that great progress is in fact being made and we know much more today than we did when we started on this journey four years ago. We are also very confident that we'll continue to accelerate this learning and make a real difference in the lives of children and young adults with TANGO2- related disease.



That is the very essence of this Conference. It highlights all of the incredible work that is currently being performed by outstanding researchers and scientists in partnership with the TANGO2 Research Foundation. It is a true collaboration between all of us – families, researchers, doctors and the foundation. This is how the best science works and how we will help our children live their best lives.

The TANGO2 Research Foundation is committed to continuing its support for these types of efforts and is currently funding a new round of projects for 2022. Thank you to all the families, researchers, doctors, board members, donors and volunteers that have made this work possible. Together, we are making a difference.

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KASHA MORRIS REGIONAL COORDINATOR PROGRAM EAST HADDAM, CT USA



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Audience Q&A

Slido is an easy-to-use Q&A platform for events that gives everyone a chance to ask their questions. You can ask anonymously and vote for the questions you like, bringing the most important topics to light. Use this QR code to get started now. 2018 The Tango2 Research Foundation is created to bridge the gap between patient/families & medical community

SFAMILIES.

2018 1st TANG02 Mouse Model was conducted at BCM. Mouse model study can help to uncover a better understanding of the basic biology of the TANG02 gene. 2019 PCORI Grant- Patient Centered Outcomes Research Institute was secured to fund a research conference.

FAMILIES

2020 NHS provides insight into prevention of life threatening metabolic crisis enabling us to create an emergency protocol letter for families to get life saving emergency care.

2019 PCORI Grant- Patient Centered Outcomes Research Institute was secured to fund a research conference.

2018 Natural History Study (NHS) conducted at Baylor College of Medicine. The study collects health information over time to understand how the medical condition or disease develops and to give insight into how it might be treated. **2020** The Foundation was awarded a \$450,00 grant from the Chan Zuckerberg Initiative. The grant allowed the Foundation to greatly accelerate the expansion of its research network and capacity building initiatives. 2020 Launched a Regional Support Program. Eleven Regional Support Coordinators volunteered to share the latest resources and information to TANGO2 families and to promote connections among TANGO2 families that live in the same region.

2020 €

2020 1st Grant Cycle funded 5 year-long research projects around the world 2021 Conducted disease prevalence study an important decision- making factor used by researchers and pharmaceutical companies to determine goals and resource allocation toward the condition. **2021** Launched new partnership with families & researchers in a rare disease registry. This partnership will bring families and researchers together to gain a better understanding of TANG02 disease and accelerate research.

Our . mission:

To improve the lives of children and young adults affected by TANGO2 related disease through helping to fund, coordinate and guide scientific research that leads to a better understanding of how TANGO2 mutations affect them.

Our goals:



Fund more research that leads to life-saving treatments.

Identify more undiagnosed patients and shorten diagnostic odyssey



Identify potential treatments for disease management



Provide support for families and children living with TANGO2 disease

2021 1st Community-Wide Fundraising Campaign Exceeds \$70,000 goal

2021 Tango2 was added to Gene Panels to help shorten the diagnostic odyssey.

2021 Presented the first TANG02 Research Symposium for families, researchers, doctors, etc. to share the latest research information and broaden our research network. (At no cost to families.)

TANGO2RESEARCH FOUNDATION



DR. SEEMA LALANI

Dr. Lalani is a Professor in the department of Molecular and Human Genetics at Baylor College of Medicine. She completed her residency in Pediatrics at Hershey Medical Center in Pennsylvania and then followship in Clinical Genetics at Baylor College of Medicine. She subsequently completed training in Clinical Cytogenetics and Molecular Genetics. Her work has focused on understanding the molecular basis of neurodevelopmental and cardiovascular disorders in children. She has also been involved with rapid exome sequencing of children with birth defects in neonatal and pediatric intensive care units. She has been involved with the Undiagnosed Diseases Network (UDN) study at Baylor College of Medicine for several years and helped characterize diagnoses in multiple undiagnosed children with rare diseases. Her team has made several novel gene discoveries related to pediatric disorders in the past decade. In 2016, her group identified the first cohort of patients with TANGO2 disorder, which led to a rewarding association with the TANGO2 Research Foundation. She currently serves as a Board member of the foundation.



DR. ERICA LAY

Dr. Erica Lay is a clinical research fellow and clinical instructor in the department of Molecular and Human Genetics at Baylor College of Medicine. She completed medical school and pediatric-medical genetics residency at Baylor College of Medicine as well. Her time during her research fellowship has focused on describing genetic conditions with dilated cardiomyopathy like LMOD2-associated dilated cardiomyopathy and TANGO2 disorder. After completing her research fellowship, she plans on practicing as a pediatric medical geneticist.



DR. SAM MACKENZIE

Dr. Mackenzie is a clinician-scientist at the University of Rochester focused on genebased therapies for neurogenetic diseases affecting children. He is board certified in pediatric neurology and completed a fellowship in neuromuscular medicine prior to joining the faculty at Rochester in 2021. He is interested in improving our understanding of the underlying mechanisms and neurological manifestations of TANGO2 disease as we move toward potential treatments in the future. Dr. Mackenzie is a member of the TANGO2 Foundation's Research Committee and has been involved with several projects including the addition of TANGO2 on commercial gene panels, determining the genetic frequency of pathogenic variants in TANGO2 on a population-wide scale, and developing more specific antibodies against the TANGO2 protein.



Dr. Chaya Murali is a pediatric clinical geneticist in the Department of Molecular and Human Genetics at Baylor College of Medicine in Houston, TX. She completed her pediatric genetics residency training at Children's Hospital of Philadelphia. Dr. Murali is interested in studying the quality of life of patients with genetic conditions, patient-reported outcomes, and patient-centered research. She also has a passion for creative writing.

DR. CHAYA MURALI



PR PASCALE DE Lonlay

Pr Pascale de Lonlay is a pediatrician and coordinator of reference centers of inherited metabolic diseases in France (network G2M) for children and adults. She is also a coordinator of the unit of metabolic diseases at Necker-Enfants Malades Hospital in Paris, France. Her interest in diseases of the energetic metabolism has allowed her to integrate her clinical experience, notably her observation of the importance of inflammation in the decompensation of her patients, and her skills in molecular biology, biochemistry, genetics, and cell biology.



DR. HORTENSE DE CALBIAC

Dr. Hortense de Calbiac is a postdoctoral researcher at Imagine Institute of Genetic Diseases, collaborator and soon to be full-time postdoctoral fellow of Dr. de Lonlay. She completed her PhD in 2019 at Sorbonne University under Dr. Edor Kabashi's supervision where she conducted research activities in the field of neurological and neurodegenerative disorders. In this context, she specialized in zebrafish model.



DR. CHRISTINA MIYAKE

Dr. Miyake is an Associate Professor in the Departments of Pediatrics at Texas Children's Hospital and Molecular Physiology and Biophysics at Baylor College of Medicine. She also serves as the Director of the Cardiovascular Genetics Inherited Arrhythmias Clinic. She specializes in the care of pediatric patients and families with inheritable arrhythmia disorders. Dr. Miyake is actively involved in clinical and translational research and is currently funded through the NIH. Her goal is to identify genes that cause arrhythmia disorders and improve the quality of care and outcomes among patients worldwide. She is one of the investigators at Baylor who first described the TANGO2 disorder in 2016.



DR. CLAUDIA SOLER-Alfonso

Dr. Claudia Soler-Alfonso is an assistant professor in the Department of Molecular and Human Genetics at Baylor College of Medicine in Houston, TX. She completed her combined pediatrics and genetics residency training at the University of Texas Health Science Center in Houston, TX and a fellowship in clinical biochemical genetics at the Children's Hospital of Philadelphia. She is board certified in both clinical and biochemical genetics. Her professional interests include studying metabolic disorder of glycogen, lactate, and pyruvate as well as the development of novel therapies for metabolic defects of energy generation. She manages metabolic problems in children with TANGO2 disorder.



SUNAIRA TEJPAR

Sunaira Tejpar is a Ph.D. Student at the Faculty of Education at Queen's University in Kingston, Ontario. As an educator, her research interest is in better understanding how to support students with exceptionalities in the classroom through a strengthsbased perspective. More specifically, her goal is to understand how students perceive their own exceptionalities during the identification process and the impact it has on their learning. Sunaira is interested in learning more about how educators can lean in on the expertise of parents of children with the TANGO2 deficiency to foster collaborative teaming to better support these students.



Dr. Zhang is an assistant professor of Molecular and Human Genetics, Molecular Physiology and Biophysics, and Internal Medicine at the Baylor College of Medicine. She is the chief of cardiovascular genetics clinic at Baylor College of Medicine and is board certified in clinical genetics and internal medicine.

Dr. Zhang is a physician scientist. Her laboratory studies the impact of gene regulation on the health and disease of the heart. More recently, the lab focuses on using induced pluripotent stem cell differentiated cardiomyocytes combined with genome editing tools to understand inherited cardiac diseases and translate these findings to the bedside.



DR. KEVIN GLINTON

DR. LILEI ZHANG

Dr. Glinton is an Assistant Professor in the Department of Molecular and Human Genetics at Baylor College of Medicine. He completed a residency in Pediatrics at the University of Virginia Health System and then fellowships in Clinical Genetics and Clinical Biochemical Genetics at Baylor College of Medicine. While at Baylor, Dr. Glinton's research has focused on the clinical applications of untargeted metabolomics as well as the characterization of rare neuro-metabolic disorders. Dr. Glinton is involved in several clinical trials in rare genetic disorders and enjoys educating students and patients on genetics and genomics.



Brandy Rawls-Castillo graduated Summa Cum Laude with a bachelor of science degree in Human Nutrition in 2015. She then furthered her education by receiving her Master of Science in Nutrition and Metabolism from University of Texas Medical Branch in 2016. She has been working at for the past 5 years in the metabolic clinic at Texas Children's Hospital. In 2020, she was promoted to clinical program coordinator. She is very passionate about nutrition in the treatment of inborn errors of metabolism, specifically urea cycle disorders and glycogen storage disorders.

BRANDY RAWLS-CASTILLOR, MS, RD, LD



DR. MICHAEL SACHER

Michael Sacher is a Professor in the Biology Department at Concordia University and an Adjunct Professor at McGill University in the Department of Anatomy and Cell Biology. His laboratory focuses on the mechanism of membrane transport between cellular compartments and diseases related to defects in this process, work which began as a research associate at Yale University. He discovered the TRAPP complexes in yeast and identified mammalian-specific TRAPP proteins. Using genetic, biochemical, structural and cell biological approaches, his group showed that they play pivotal roles in membrane transport and have linked mutations in a number of TRAPP genes to human disease. His laboratory recently began characterizing the function of TANGO2 using a variety of model systems, demonstrating a role in both membrane transport and at the mitochondria.



DR. LINA GONZALEZ

Dr. Lina Gonzalez is an assistant professor of pediatrics in the division of Genetics and Medical Genomics at University of Pittsburgh. She received her degree in Medicine from the University of Aleppo School of Medicine in Aleppo, Syria. She is board certified in Internal Medicine, Medical Genetics and Genomics and Medical Biochemical Genetics. Dr. Gonzalez has been awarded a National Institutes of Health (NIH) K08 Mentored Clinical Scientist Research Career Development Award under National Human Genome Research Institute (NHGRI) in 2019 for her grant "Precision Genomic Medicine in The Plain Communities and its Impact on The Plain and General Population". She also has been funded for one year from the TANGO2 Research Foundation in 2020. Dr. Gonzalez's focus is on the genetic disorder in the Plain people (Amish and Mennonites) in Western Pennsylvania to characterize novel genetic disorders or novel mutations. She also has interest in studying the pathophysiology of TANGO2 deficiency.



MIKE MORRIS AND KASHA MORRIS

Mike and Kasha Morris are co-founders of the TANGO2 Research Foundation. After more than a decade of searching, their son, Ryan, was diagnosed with TANGO2-Related Disorder in 2017. The foundation was established in 2018 to help support research for better understanding of the disorder. Mike is the creator and CEO of RecDesk, a cloud-based recreation software company and Kasha is a special education teacher. They live on a farm in East Haddam, Connecticut.



ANN GEFFEN

Heading into her third year, Ann Geffen is the TANGO2 Research Foundation's first Executive Director. She spearheads all aspects of the Foundation from foundation strategy, operations, communications, volunteer recruitment to fundraising. Geffen holds a Juris Doctor degree with the Thomas Jefferson School of Law and a Master of Arts degree in Strategic Public Relations from the University of Southern California.

She lives in Winter Garden, Florida with her husband, Dr. Joseph Geffen, and two children, Zander and Aria.



CHRIS NIKIC

Chris Nikic set a new world record by being the first person with Down Syndrome to complete a 140.6-mile Ironman. His mission is to inspire others like him to pursue their dreams and goals.

His greatest achievement is showing others how shifting your focus from your disabilities to your abilities and pursuing your dreams with an unwavering tenacity, a positive attitude and a no-quit grit can help anyone achieve their goals and dreams.



NIK NIKIC

Nik brings over 20 years of experience in helping clients maximize top line revenue by implementing the 1% Better system for sustainable continuous improvement.

Over the years Nik and his organization have worked with many global companies like United Technologies, Cognizant, United Airlines, Enterprise, Quad Graphics, Computer Sciences Corporation, NICE Systems and many others. His greatest coaching achievement: "Teaching my son how to achieve the impossible by getting 1% Better Every Day."



AMANDA & JORDAN TAGGART

Jordan and Amanda Taggart are TANGO2 parents to a 4 year old son, Jake, who was diagnosed at age two. Since then, they have tried to learn as much as they can about TANGO2-Related Disorder and how to care for a child with this disorder. Jordan leads sales for Vida Health, a virtual chronic care company, and Amanda has 15 years of experience as an elementary school teacher. As a family, they enjoy spending time together outdoors and at Lake Michigan where they live in Chicago.



SAM BURGESON

Samantha (Sam) Burgeson is the Fundraising Chair for the TANGO2 Research Foundation. She first became familiar with the Foundation when her nephew, Jake, was diagnosed with this ultra-rare genetic disease a few days after his 2nd birthday in May 2020. She lives in Chicago, USA with her husband, Jon – only a ten-minute drive from Jake & family! In her free time, Sam loves to travel & walk Chicago, finding new restaurants & enjoying the Lake Shore path. Samantha currently works in Finance within the Pharmaceutical & Health Care industries.



EBONY HILL

Ebony Hill, is a mother of two boys, Orion 7 and Osiris 5 both with TANGO2-Related Disorder. She is a full time mom and college student aiming for a career in social work. They live just outside Toledo Ohio.



VERONICA JONES

Veronica is the mother of Thea, a 7-year-old child with TANGO2-Related Disorder. She is a first-grade teacher and enjoys reading, yoga, and gardening. Her and her family live in San Diego, California.



Amanda Hull is from Suffolk in England. Her and husband, Daniel, have three boys-Joe 13, Sebby 11 and Walter 6. Sebby has TANGO2-Related Disorder. They live on a farm and have an active lifestyle with all of them enjoying being outdoors and doing adventurous activities. Amanda is an Educational Psychologist and works in schools, and Daniel is a farmer.

AMANDA HULL

Conference Agenda

Sunday, June 26

5:00 - 7:00 PM | Welcome Reception

Monday, June 27

7:00 - 9:00 AM | Breakfast 9:00 - 9:15 AM | Welcome to Day 1 with Kasha Morris 9:15 - 10:00 AM | Keynote Speakers Chris & Nik Nikic - 1% Better 10:00 - 10:15 AM | Faces of TANGO2 disease 10:15 - 10:45 AM | TANGO2-Related Disorder 101 Dr. Erica Lay Break 11:00 - 11:30 AM| Building our Understanding of TANGO2-Related Disorder From Gene to Child Dr. Samuel Mackenzie 11:30 AM - 12:00 PM | Mitochondrial Dysfunction in TANGO2-Related Disorder; What is next? Dr. Lina Gonzalez 12:00 - 12:30 PM | Video: Assessment of Metabolic Studies in TANGO2-Related Disorder Drs. Claudia Soler and Kevin Glinton 12:30 - 1:55 PM | Lunch 2:00 - 3:30 PM | Workshop: What matters most to families? Amanda & Jordan Taggart, Dr. Sam Mackenzie Break 3:45 - 4:15 PM | TANGO2-Related Disorder Natural History Study – How families are helping to save lives Dr. Christina Miyake 4:15 - 4:45 PM | It takes two to tango: the search for partners for TANGO2 Dr. Michael Sacher *Please note, schedule is subject to change

Conference Agenda

Tuesday, June 28

7:00 - 9:00 AM Breakfast
8:00 - 9:00 AM Science Meeting, Committee Recruitment, & Siblings Meetup
9:00 - 9:15 AM Welcome to Day 2 with Dr. Seema Lalani
9:15 - 9:45 AM About the TANGO2 Research Foundation with Kasha & Mike Morris
9:45 - 10:15 AM Cardiac Crises – What physicians and families need to know
Dr. Christina Miyake
Break
10:30 AM - 11:45 PM Breakout Sessions:
Parenting Tips- Ebony Hill & Amanda Hull
Nutrion & TANGO2-Related Disorder - Brandy Rawls-Castillo
Educational Needs of individuals with TANGO2-Related Disorder -
Sunaira Tejpar & Veronica Jones
12:00 - 1:25 PM Shine a Light on TANGO2 Campaign Celebration Lunch
1:30 -2:00 PM Family perspectives and priorities in TANGO2-Related Disorder: Using
patient-centered approaches to guide clinical care and research
Dr. Chaya Murali
2:00 -2:30 PM Video: Drug screening for Tango deficiency and emergency certificate
Pr. Pascale De Lonlay and Hortense de Calbiac
2:30 - 3:00 PM Lessons from beating heart cells in the dish on TANGO2 arrhythmia
Dr. Lilei Zhang
Break
3:15 - 4:30 PM Questions & Answers with Dr. Seema Lalani

Conference Expibi

During breaks and lunch, please take a moment to visit our exhibits

Poster Presentations

We invite you to view posters highlighting the TANGO2-related studies by our physicians, scientists, and students at the conference.

Note: Posters are to be displayed before break on Monday and removed after lunch Tuesday.

Located at Coronado M

BioBank

DNA samples are being collected (blood, saliva, skin) as part of the TANGO2 Natural History study. If you haven't had a chance to participate yet or would like more information, please see Alicia Turner and Dr. Mahshid Azamian at the biobank station.

Located at Registration

Monday: 8-9 am & 12:30 - 2:00 PM Tuesday: 8-9 am & 5:00 - 6:00 PM

CoRDS & Natural History Study Sign Up

Patients with TANGO2 disease are invited to participate in the Natural History Study (NHS) led by Dr. Christina Miyake during breaks and lunch to pick up information. Please see Dr. Mahshid Azamian.

Located at Registration

Merchandise

Suport our Foundation by purchasing a calendar or conference t-shirt.

Located at Registration





TANGO2	TANGO2 is a gene responsible for performing a specific job in the body which is actually the acronym it stands for Transport And Golgi organization.						
Golgi	Golgi functions as a factory in which proteins received from the endoplasmic reticulum are processed and sorted for transport to their eventual destinations.						
Endoplasmic Reticulum	Endoplasmic reticulum is a collection of tubes that make, package, and transport proteins and fats.						
	Likened to the cell's "post office," the Golgi apparatus modifies, sorts and packages proteins for secretion.						
Exons	An exon is a coding region of a gene that contains the information required to encode a protein.						
Deletion	A genetic deletion means that a part of a chromosome is missing. A very small piece of a chromosome can contain many different genes. When genes are missing, there may be errors in the development of a baby, since some of the "instructions" are missing.						
Misspelling	A misspelling is a change or mutation in a gene.						





This is a condition inherited from both parents that results from having no functioning copies of a gene.

Mitochondria are the parts of our cells that generate energy from food that the rest of the cell can use.

Metabolism is all the chemical reactions involved in converting food into energy.

Metabolomics metabolites, within cells, biofluids, tissues or organisms.

Recurrent Metabolic Crises

Autosomal

Recessive

Mitochondria

Metabolism

Our body has an order or a way that it works best to give us energy and keep us healthy. When the TANGO2 gene doesn't work in the body, it disrupts this order and it causes a crisis because we can't utilize this typical way of making energy. We can measure metabolic crisis with lab tests. The body uses up a lot of its sugar resulting in low blood sugar which leads to hypoglycemia. There can be a build up in lactic acid called lactic acidosis. Lactate and glucose are closely related and so a decrease in one can cause an increase in another. We can also see elevated ammonia called hyperammonemia.

Enzymes

An enzyme is a protein molecule in cells that speeds up chemical reactions in the body, but does not get used up in the process. Therefore it can be used over and over again.



Fatty Acid Oxidation	During digestion the body breaks down fats into fatty acids which can then be absorbed into the blood. Fatty acids have many important functions in the body, including energy storage. If glucose (a type of sugar) isn't available for energy, the body uses fatty acids to fuel the cells instead.						
Membrane Traffic	Membrane trafficking is the process by which proteins and other macromolecules are distributed throughout the cell.						
Carnitine	Carnitine is a natural substance that the body uses to process fats and produce energy. Carnitine deficiency is when not enough (less than 10%) of the nutrient carnitine is available to cells in the body. This can cause muscle weakness and heart or liver problems.						
Rhabdomyolysis	This is the breakdown of muscle. With TANGO2 there could be muscle injury from all the abnormal labs of a metabolic crisis that cause the muscle fibers to die and break down Muscle that breaks down releases myoglobins. A kidney that is overwhelmed with myoglobin can be injured. We measure how much muscle is breaking down by measuring the transfer of Creatine kinase or CK.						
	IURN RUSLEL MIRLINNA VALENS RUNDSHEM URINE						

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Myoglobins	Myoglobin is a protein found in heart tissue and other muscles. It is released into the blood after damage to the heart or other muscles. It can be checked with a blood test or a urine test.							
Cardiac Arrhythmias	An arrhythmia is any change in heart rhythm. Hearts typically have a very specific wave and the PQRST describes the normal shape of that wave. With arrhythmia related to TANGO2, the most common change is an increase in space between the Q and T part of the wave called QT prolongation. Any change to our normal heart wave is dangerous because it may not snap back into normal rhythm and the heart can stop.							
Tachycardia	Tachycardia is an abnormally fast heart rate.							
Cardiomyopathy	Cardiomyopathy is when the heart muscle becomes weak and enlarged, which makes it difficult to pump blood through the body.							
	Normal heart rhythm Irregular heart rhythm							
	-Anton Anton							

Myocytes

These are a type of cell found in muscle tissue. They develop from myoblasts to form muscles in a process called myogenesis.

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En	ce	ph	al	op	at	hv
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This is damage to the brain that changes the firing patterns of the mind. Our brain activity is measured in waves. An EEG is an electroencephalogram which measures the electrical impulses of the brain. EEGs are supposed to show minimal peaks but with encephalopathy, there are peaks and lows that don't follow a normal pattern. This is an indication that the brain is not functioning correctly. This may explain delays and issues in learning in some children.

BiomarkerThis is a biological molecule found in blood, other body
fluids, or tissues that is a sign of a normal or abnormal
process, or of a condition or disease. A biomarker may be
used to see how well the body responds to a treatment
for a disease or condition.

Pluripotent Stem Cells (iPSC)

Fibroblast

Pathophysiology

CRISPR

These cells are master cells. They are able to make cells from all three basic body layers, so they can potentially produce any cell or tissue the body needs to repair itself. This "master" property is called pluripotency.

Skin fibroblast cells are often very important for research analysis. These cells can help us understand the function of the TANGO2 gene.

This is changes in the body associated with a particular disease or injury, or the study of such changes.

CRISPR is a technology that can be used to edit genes.



OUR IMPACT

SINCE OUR INCEPTION IN 2017, WE'VE MADE GREAT STRIDES TOWARD FINDING TREATMENTS AND A CURE FOR TANGO2 DISEASE.





Why is skin biopsy done?

Skin biopsy is often done in children suspected to have rare genetic diseases. Skin biopsy is a procedure often performed in children by pediatric geneticists to help gain understanding of the genetic disorder that they have. Skin is made up of cells called fibroblasts. Once skin biopsy is done, the fibroblasts are grown in a special nutrient medium to get millions of copies. These cells can then be studied to determine which cell function is affected by the specific genetic change that the child has. Many times, for new disease genes, such as TANGO2, not enough is known about what goes wrong in cells when the gene is not working. Are the cells not able to produce energy? Is that the reason children develop muscle or heart problems? Are the cells not able to utilize glucose or other nutrients when stressed? What stresses the cells? Importantly, are there medications that can improve the way cells function despite having the gene mutation? Could these be the medications that can be used to treat children? Skin fibroblasts can help us learn a lot about what TANGO2 gene does, and why children develop problems when the gene is not working. As we understand more about he workings of TANGO2, we can hopefully develop ways to treat this disease in the future.

How is skin biopsy done?

First, a numbing cream (called EMLA) is applied to the inside of the arm for about 30 minutes. This provides enough time for the skin area to become numb. Then, another numbing medication, called lidocaine is injected at the same spot, using a fine needle to ensure further numbing of the skin. After the skin is well-anesthetized, a 2 mm punch biopsy is obtained. This small piece of skin is then put in a pink liquid (culture medium). The site is bandaged which can be removed the next day. The procedure takes about 5-10 minutes, excluding the EMLA cream application time. The skin can be cleaned with soap and water routinely after the bandage is removed.

The skin sample is taken to a research lab where it can be grown. It can take several weeks for skin fibroblasts to grow to an adequate number of cells. Once expanded, these cells can be studied directly in a laboratory or frozen in a freezer for future studies.

Please contact Mahshid Azamian at azamian@bcm.edu or Seema Lalani at seemal@bcm.edu for further questions.



Image source: https://www.podiatrytoday.com/guide-biopsy-techniques

CORDS Rare Disease Registry

The TANGO2 Research Foundation has partnered with Coordination of Rare Diseases at Sanford (CoRDS) to house patient registry data. CoRDS is a centralized international patient registry for rare diseases based at Sanford Research, a nonprofit research institution.

HOW DOES CORDS COORDINATE THE ADVANCEMENT OF RESEARCH FOR 7,000 RARE DISEASES?

- Works with patient advocacy groups, individuals and researchers.
- Captures health information from individuals with a rare diagnosis, undiagnosed patients, unaffected carriers or at-risk patients.
- Connects researchers and patients and notifies participants of emerging clinical trials.
- Makes the registry accessible. Participants can enroll for free and researchers can access it for free.

PLEASE ENROLL TO HELP RESEARCHERS IDENTIFY EMERGING CLINICAL TRIALS FOR TANGO2 DISEASE PATIENTS TANGO2RESEARCH.ORG/PATIENT-REGISTRY

TANGO2 DISEASE

NATURAL HISTORY STUDY

Baylor College of Medicine

PLEASE ENROLL TO HELP IDENTIFY NEW TREATMENTS AND THERAPIES FOR TANGO2 DISEASE PATIENTS

TANGO2RESEARCH.ORG/TANGO2-STUDY-NOW-UNDERWAY

Researchers at Baylor College of Medicine invite you to participate in the TANGO2 Natural History Study & **THEY NEED ALL TANGO2 DISEASE PATIENTS TO ENROLL!** This study can help understand the progression of disease over time, provide opportunities to improve clinical care, and establish treatment guidelines. It is crucial that we have the medical information from all affected families. For more information, email Dr. Mahshid Azamian at tango2.research@bcm.edu.

Calling all families!





www.tango2research.org

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