**OpenMinds Clinical Packages/AUBMC Special Kids Clinic (ASKC)/Research**

**For the ASKC and OpenMinds the aforementioned financial, health and other calamities facing Lebanon have only translated into a larger than ever number of children with special needs requiring diagnosis, management and assistance at all levels.**

**Since inception of Openminds the number of children receiving support has increased to 573 families as of August, 2022 (40 in 2014) representing 93% growth over 7.5 years!**
**This excludes the number of families requiring augmented/additional assistance the past three years: 39% increase in 2020; 44% increase in 2021; 50% increase in 2022.**

**The increase in therapy hours provided by the ASKC to children and adolescents increased by >50% from January to August 2021 to 2022. This has necessitated recruitment of pediatric neurology physicians from two to four highly qualified individuals and a need for additional therapists in all disciplines, which will translate into expansion in space to provide vital services.**

***We also continue to speak for those who cannot by our social, national and international media presence. Examples from late 2021 and 2022 are:***
*Sep 12, '21. PODCAST:* ***Professors at Work*** *(Rami Khoury): She leads genes, cells, and molecules into battle against rare childhood diseases. American University of Beirut: Rose-Mary Boustany.* ***she-leads-genes-cells-and-molecules-into-battle-against-rare.mp3***
*March 9, '22. Faculty of Health Sciences-AUB:* ***Autism in Lebanon: 2022***
*March 11, '22.* ***THE RESEARCHER****. LBC. Season 2-Episode 5 (****autism****). https://youtu.be/Jue5DQR1buo.*
*S Saab, J Makoukji, K Kanaan, G Hassan, N Makhoul, J Soueid, P Trippier, R-M. Boustany.* ***Flupirtine Derivatives as Treatment for CLN6 Disease*** *(Poster-May 9/10 '22). Research of the Future: Challenges and Opportunities (AUB-6th CRSL Research Conference. Kamal Shair Auditorium AUB).*
*Jihane Soueid, Lara Al-Bazzal, Rose-Mary Boustany.* ***Novel Autism Genes and Perturbed Proteastasis. AUB-6'th CRSL Research Conference). May 10, 2022.*** *Research of the Future: Challenges and Opportunities (AUB-6th CRSL Research Conference. Kamal Shair Auditorium).*
*Sara Saab, Joelle Makoukji, Karen Kanaan, Rose-Mary Boustany Batten Disease Support and Research Association Virtual Meeting U.S.A:* **Phenotyping of the Cln6 or nclf mouse model: Retinal, behavioral and molecular aspects.** July 9th 2022.

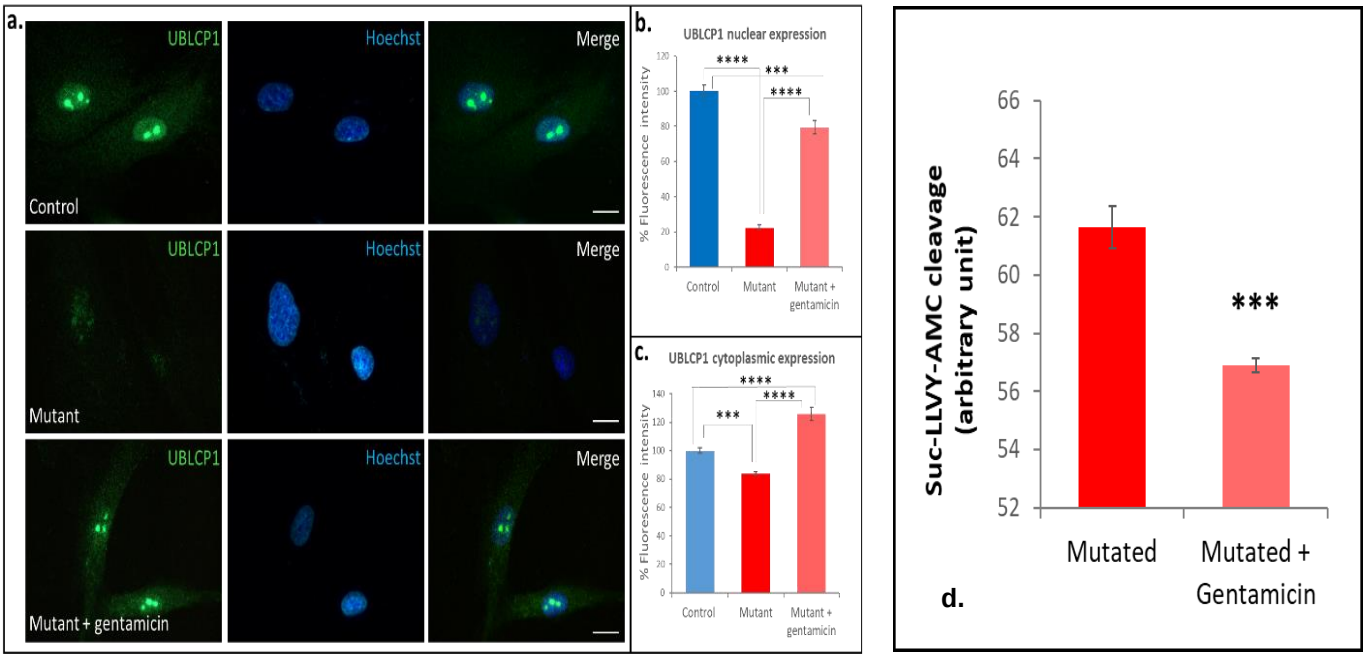
**Thanks to crucial funding provided by OpenMinds to the ASKC and the Neurogenetics unit academic productivity has soared with international publications increasing to 116 from 68.**

***Progress that translates to help for complex neurodevelopmental conditions, like autism, takes time, effort and funding. We provide care for more than 1000 families with children with autism at the ASKC. We began in 2010 by collecting blood samples from 41 families, establishing prevalence, publishing on prevalence, risk factors and positive impact of crucial early intervention on the brain and clinical outcome in autism. We have now identified 4 novel genetic defects causing autism.***

## *For one of those defects, 12 years later, we have a handle on a possible therapy*

A Lebanese family with autism or ASD has a defect in the **UBLCP1 gene** consisting of a **premature stop** resulting in a shortened and less stable UBLCP1 protein. The normal function of the UBLCP1 protein is to dampen protein cell breakdown. When UBLCP1 is diminished or non-functional, protein breakdown becomes out of control resulting in cell damage. Gentamycin or the safer ataluren are drugs that can trick cells into reading across the premature stop helping them increase amount of UBLCP1 protein and restore function.


**UBLCP1 gene with premature stop**




**Amount of UBLCP1 and function are restored in skin cells from the ASD patient after treatment with gentamicin.** (**a**) Normal skin cell (upper panel) and cells with UBLCP1 ASD mutation (middle panels) treated with gentamicin (lower panels), immune-stained for UBLCP1 (green) and nucleus dye (blue). (**b and c**) UBLCP1 nuclear and cytoplasmic protein expression are low in cells from the ASD patient. Note: restoration of UBLCP1 protein after gentamycin treatment. (**d.**) **UBLCP1 function is restored in mutant/ ASD cells by gentamicin.** A desired decrease in proteasome activity occurs in cells from ASD patient after treatment.