PURA Syndrome Foundation awards Fellowship Grant

Clinical research fellow position at University of Southampton to establish and administer the Global Patient Registry, undertake research into the Natural History of PURA syndrome and publish future papers.

Tulsa, Oklahoma, USA—June 2018

The PURA Syndrome Foundation announced that it has awarded a research grant to the University of Southampton, in the United Kingdom. The £20,000 (approx. $26,700) grant directly fulfils a portion of the foundation's mission to further research into this rare genetic condition. The funds were raised by families and friends affected by the PURA syndrome.

This research grant will help fund an 18-month position at the University of Southampton. This role is jointly funded by the Foundation and by an anonymous donor to the University of Southampton, UK. The successful applicant will help develop and administer a secure global online patient registry. Information from this registry will form the basis of future research papers and help us to better understand what happens with PURA syndrome across the life span of patients (longitudinal study), creating a Natural History of the syndrome.

Both the study and Fellow will be overseen by the grant applicant, Prof Dr Diana Baralle. Dr Baralle holds a prestigious NIHR Research Professorship. As a Professor of Genomic Medicine and Clinical Geneticist, Prof Dr Diana Baralle's research spans clinical phenotyping, diagnostics, and molecular mechanisms. Her clinic provides a rich resource for genetics and genomics research finding new causes for rare disorders. Dr Baralle's laboratory takes this further by studying the molecular mechanisms in pre mRNA splicing which will helps them improve diagnostic testing, understand how genes work and therefore how they can modify them for therapy.

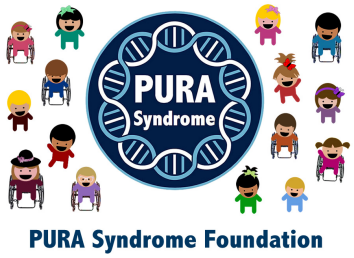
"This grant is a great accomplishment for the foundation and will provide an exciting next step in PURA syndrome research and understanding for our families" said Dominic J. Spadafore, the President of the Foundation. "We expect that this will be the first of many grants."

PURA syndrome is a rare genetic disorder, primarily associated with neurodevelopmental abnormalities. The PURA gene is located on the long arm of chromosome 5 (at 5q31.3). PURA is expressed in all tissues, including the brain, muscle, heart, and blood.

PURA syndrome was first described in medical literature in 2014. As of June 2018 there were at least 250 individuals diagnosed with the condition globally. With the increasing use of the latest 'gene sequencing' technology, it is expected that more individuals, both children and adults, will be diagnosed with this condition over the next few years.

Foundation Logo
Photo courtesy of the PURA Syndrome Foundation.

Research Lab, University of Southampton UK.
Photo courtesy of the PURA Syndrome Foundation.



PURA kids
Photo courtesy of the PURA Syndrome Foundation.

All individuals with PURA syndrome have moderate to severe intellectual disability. Most individuals remain non-verbal, with severe language and motor delay. However, many have better receptive language than expressive language. Over half of the individuals have been diagnosed with epilepsy.

The clinical fellowship position has been awarded and the successful applicant is Dr Rebecca Mawby. Dr Mawby is a trainee Paediatrician with experience in complex paediatric disorders. She is continuing study in the area of epilepsy and seizures, and endocrinology, which will further benefit the PURA syndrome community. Dr Mawby will assist in the development of the global patient registry, completing research study from information gathered. She is expected to start early in September 2018.

About the PURA Syndrome Foundation

The PURA Syndrome Foundation is a global organization, registered in the USA as a 501(c)(3) tax-exempt non-profit corporation. The PURA Syndrome Foundation supports and educates patients and their families, providing a global community. This community provides a place of belonging to those who may otherwise feel isolated by rare disease, enriches the medical research being completed and educates those outside the community about the condition. Their mission is to serve, educate and fund research for families coping with the effects of PURA syndrome. The foundation was established in 2016 and is registered in the United States. The Foundation has board members in the EU, Australia, and the USA. The Foundation strives to bring together and support families across the globe. To learn more, visit **www.purasyndrome.org/**

Related Resources:

PURA Syndrome Foundation announces award of Fellowship https://www.purasyndrome.org/single-post/2017/11/21/Together-as-a-global-community

PURA Syndrome Foundation Research News – New PURA syndrome clinical paper published https://www.purasyndrome.org/research

Related Link:

PURA Syndrome Foundation: www.purasyndrome.org

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