The PURA Global Patient Registry: Understanding PURA Syndrome

We encourage all families to participate in this long-term study to develop a better understanding of PURA syndrome and how it affects people over time. Participation requires the online completion of several questionnaires covering a wide range and depth of information relating to the individual with PURA Syndrome.

The questionnaires do not all have to be completed at once. However you will need to gather some medical information or have access to medical records in order to complete some of the sections. It will also be necessary to add some reports to the online system, especially those confirming the PURA diagnosis and specific mutation. Please see the "Uploading Documents" section below for instructions on how to do this.

To help you prepare for the completion of the questionnaires we have provided a checklist below to indicate the scope of questions to be answered and the sorts of information which will be required. *There are a few sections that would be extremely helpful if you prioritized entering the information into them first. They are indicated with *** on the checklist.*

Thank you for your involvement! The study team appreciates the investment of time and effort required to comprehensively complete the questionnaires. Completing this study will give our researchers a better understanding of PURA syndrome and how it affects people over time.

Checklist of items you will need for each Module
*****indicates priority sections - please fill out first*****

Participant Information: Demography Participant***

- ☐ Basic information: name, address, contact, person entering information
- ☐ Genetic Diagnosis
- ☐ Recommended Clinicians with contact information

Module 1: Genetics and Family History***

This section investigates any genetically tested and confirmed PURA mutation of your child, both parents, siblings and the wider family.

- ☐ Any diagnoses of developmental delay, autism, epilepsy or congenital abnormality amongst family members
- ☐ Family history of miscarriage, stillbirth or the deaths of babies and children

Module 2: Pregnancy, Birth and Newborn***

Explores a range of events during pregnancy including movement and position of the baby, maternal illnesses such as high blood pressure, anaemia, gestational diabetes or pre-existing maternal medical conditions.

- ☐ Ages of both parents at conception date
- ☐ Maternal use of medications pre-conception and during pregnancy
- ☐ Length of pregnancy, natural or induced labour, type of delivery, complications during delivery, APGAR scores for baby after the birth
- ☐ Birth weight, head circumference and length
- ☐ Abnormalities noted in the neonatal period (these are listed for your assistance).
- ☐ Newborn feeding including by naso-gastric tube or intravenously
- ☐ Any admission to a neonatal, high dependency, special care or paediatric unit within the four weeks after birth

Module 3: Developmental Milestones (Part 1)***

- ☐ Current Measurements - age, weight, head circumference, length/height, intellectual or learning disability
- ☐ Gross motor skills e.g. lifting head, rolling, sitting with/without support, walking
- ☐ Fine motor skills e.g. eyes fix and follow objects or people, grabs onto objects within reach, passing objects from one hand to the other
- ☐ Eye sight issues and treatment

Module 3: Developmental Milestones (Part 2)***

Social Skills

- ☐ First smile, giggle/laugh, recognition of familiar faces
- ☐ Self feeding with hands, drinking from a beaker, use of fork/spoon
- ☐ Toilet training
- ☐ Waving hello/goodbye, clapping hands
- ☐ Pointing or gesture to a wanted object or item
- ☐ Types of interaction with others e.g. plays simple games, chooses or has preference for a certain friend
- ☐ Eye contact
- ☐ Separation anxiety, showing fear or aggression

Communication (spoken and understanding)

- ☐ Startles at loud/sudden noises
- ☐ Turns head/eyes towards sounds and voices
- ☐ Gurgling or cooing, babble, first word, vocabulary, linking words, making sentences or conversation
- ☐ Responds to name, understands 'no'
- ☐ Follows one-step command or two-step command
- ☐ Communication type e.g. gestures, facial expression, assistive technology
- ☐ Hearing issues

Module 4: Neurology***

- ☐ Abnormal movement or movement disorder
- ☐ Repetitive hand movements e.g. hand wringing
- ☐ Abnormal blinking
- ☐ Limb shaking or tremors
- ☐ Sweating, temperature regulation (too hot, too cold)
- ☐ Circulatory issues e.g. cold hands/feet
- ☐ Excessive sleepiness
- ☐ Low or floppy muscle tone, High or stiff muscle tone
- ☐ Weakness in muscles of arms or legs
- ☐ Responds to pain
- ☐ Appearance of feet - descriptions are provided for your assistance
- ☐ Responds to smell/strong odours
- ☐ Blinking in response to bright light
- ☐ Variations in facial expression
- ☐ Symmetrical facial movement
- ☐ Gag reflex
- ☐ Ability to turn head from side to side
- ☐ Can your child poke their tongue out
- ☐ Safe swallow
- ☐ MRI or CT scans of brain
- ☐ Electroencephalogram /EEG
- ☐ Electromyography/EMG

Module 5: Seizures and Epilepsy

- ☐ Age at first seizure, association with high temperature
- ☐ Diagnosis of epilepsy, type of seizures, triggers, warnings or changes in behaviour pre-seizure
- ☐ Time of seizures e.g. morning/evening/night
- ☐ Frequency, apparent factors affecting frequency or clustering of seizures
- ☐ Duration of seizures and how they usually stop/terminate
- ☐ Recovery time
- ☐ Successful seizure medications used including in emergency treatment - medications are listed for your assistance
- ☐ Unsuccessful seizure medications tried, including those stopped due to side effects - medications are listed for your assistance
- ☐ Ketogenic diet use
- ☐ Vagal nerve stimulator
- ☐ Changes in development since onset of seizures

Module 6: Growth

Includes mother and father's height, whether your child was born prematurely and your child's age, head circumference, height and weight prior to joining the study.

Module 7: Endocrine

- ☐ Bone health
- ☐ Thyroid and/or pituitary function
- ☐ Scoliosis, hip dysplasia or other hip problems
- ☐ Teeth problems
- ☐ Broken/fractured bones, low bone density, thin or brittle bones, osteoporosis/osteopenia
- ☐ DEXA or bone mineral density scans
- ☐ Low Vitamin D or calcium levels
- ☐ Any supplements given
- ☐ Renal stones
- ☐ Puberty - age over 8 years/signs

Uploading Documents

- Within the patient participant screen go to “Dossier options” (button, bottom left of screen)
- Select ‘Runtime data pdfs’
- In the top right of the page select “upload pdfs”
- The PDF name will need to be in the format “Subject ID_ Event ID” for example, if your subject ID is PURA0034, your file name will need to be “PURA0036 _MODUPAEN” for the PDF to be attached to PURA0036 Participant Modules events.

Clinician Involvement

If you think it would be beneficial that your clinician joins the study in relation to your individual with PURA Syndrome, we request that you first discuss the study with your clinician and obtain their consent. Then, you may provide their contact details to the Southampton University study team (PURA@soton.ac.uk), who will then contact the clinician directly so that the clinician and your participant questionnaires can be linked on the system.