



Tools and Resources for PURA Syndrome Researchers and Clinicians

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This resource provides a comprehensive overview of key tools and data sources to support PURA Syndrome research and clinical work. This centralized resource is designed to facilitate collaboration and accelerate research and therapeutic development for PURA Syndrome. Information was gathered and formatted by [Odylia Therapeutics](#) and the [PURA Syndrome Foundation](#).

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Biobanks

Source Country	Description
Germany	<p>The PURA-Biobank will involve the collection of a combination of prospective and retrospective samples from consented PURA syndrome patients globally. The samples will be used for research with the long-term goal to understand and develop treatment strategies against PURA syndrome or related disorders that include genomic deletion or duplication of the PURA gene as well as other neurological disorders.</p>
Australia	<p>The Australia Biobank is part of a large research program focused on developmental and epileptic encephalopathies, including PURA Syndrome. The program aims to identify new genetic causes, characterize over 900 known disorders, and explore their psychosocial and health economic impacts. It supports the development of new standards of care and prepares for precision medicine trials. The biobank collects specimens from participants, links clinical information to cellular studies, and generates cell lines to investigate disease mechanisms.</p>
USA	<p>The COMBINEDBrain Biorepository is a collaborative initiative aimed at accelerating research into rare genetic neurodevelopmental disorders. Managed by COMBINEDBrain, a nonprofit consortium of over 60 patient advocacy groups, the biorepository collects and stores biological samples—such as blood, urine, and tissue—from individuals diagnosed with these conditions. These samples are made available to researchers worldwide to identify biomarkers and develop targeted treatments.</p>

Patient Data

Data type	Institution/ Organization	Contact	Description
Natural History Study	University of Southampton	D.Baralle@soton.ac.uk	The PURA Syndrome Longitudinal Natural History Study is an ongoing, web-based registry launched in 2022 to collect comprehensive clinical data from patients with PURA syndrome to understand its long-term progression, management, and outcomes. Led by the University of Southampton , the registry allows families and clinicians to contribute data on the condition's symptoms, development, and overall health, providing essential information for managing the rare genetic disorder and supporting drug development
Patient Registry	Managed by the PURA Syndrome Foundation through COMBINEDBrain	PURA Syndrome Foundation	The COMBINEDBrain Patient Registry is a global initiative designed to accelerate research and treatment development for rare genetic neurodevelopmental disorders. Managed by COMBINEDBrain, a nonprofit consortium of over 60 patient advocacy groups, the registry collects and centralizes health data from individuals diagnosed with these conditions.

Research Models

(*): Unknown (?): Needs to verified (-): Does not exist

Cell Models

Cell type	Genotype	PI/ Institution	Availability/Contact	Publications
Patient-derived [unknown cell type]*	*	James Dowling, MD, PhD	James.Dowling@penmedicine.upenn.edu	-
iPSC	PURA_A89P (+/+, +/-, -/-)	JAX	Isogenic control (+/+) Heterozygous knockout (+/-) Homozygous knockout (-/-)	-
iPSC	PURA_F271del (+/+, +/-, -/-)	JAX	Isogenic control (+/+) Heterozygous knockout (+/-) Homozygous knockout (-/-)	-
iPSC	PURA_G179 (+/+, +/-, -/-)	JAX	Isogenic control (+/+) Heterozygous knockout (+/-) Homozygous knockout (-/-)	-
iPSC	PURA KO (-/-)	Dierk Niessing, PhD	dierk.niessing@helmholtz-munich.de	(Bacher et al. 2025)
*	*	Christopher Reid, Florey Institute, Australia	christopher.reid@unimelb.edu.au	-

Mouse Models

Species	Genotype	PI/ Institution	Availability/Contact info	Publications
C57/BL6	PURA (+/-), PURA (-/-)	Jennifer Gordon, Temple University		(Barbe et al., 2016) (Mishra et al., 2013) (Johnson et al., 2006) (Khalili et al., 2003)
BALB/c	PURA (-/-)	Jochen Herms, PhD, DZNE	jochen.herms(at)d zne.de https://www.informatics.jax.org/allele/allgenoviews/MGI:5305287#MGI:5305318	(Hokkanen et al., 2012)
*	PURA KO (?)	Dierk Niessing, PhD	dierk.niessing@he lmholtz-munich.de	-
*	PURA KI (p.Phe233del)	Christopher Reid, Florey Institute, Australia	christopher.reid@u nimelb.edu.au	-
C57BL/6J	PURA (-/-) exon 2 deleted	JAX	C57BL/6J-Puraem2Lutzy/Mmjax	-
C57BL/6J	PURA cKO exon 2 deleted	JAX	C57BL/6J-Puraem 1Lutzy/Mmjax	-
*	*	Stanley lyudurai		-

Frog Models

Species	Genotype	PI/ Institution	Availability/Contact info	Publications
<i>Xenopus</i>	PURA KO	Matt Guile, PhD, University of Portsmouth	Matthew.Guille@p ort.ac.uk	-

Zebrafish Models

Species	Genotype	PI/ Institution	Availability/Contact info	Publications
<i>Danio rerio</i>	<i>puraa</i> ^Δ <i>mde28</i> Gt ^Δ	Bettina Schmid, DZNE	https://zfin.org/ZD/B-ALT-250402-2#summary https://www.ezrc.kit.edu/	-
<i>Danio rerio</i>	<i>purab</i> ^Δ <i>mde29</i> Gt ^Δ	Bettina Schmid, DZNE	https://zfin.org/ZD/B-ALT-250415-1	-
<i>Danio rerio</i>	<i>Puraa</i> (-/-); <i>purab</i> (-/-)	James Dowling, MD, PhD	James.Dowling@penmedicine.upenn.edu	Data presented at 2024 PURA Syndrome Conference

Worm Models

Species	Genotype	PI/ Institution	Availability/Contact info	Publications
<i>C. elegans</i>	<i>plp-1</i> (-/-)	James Dowling, MD, PhD	James.Dowling@penmedicine.upenn.edu	Data presented at 2024 PURA Syndrome Conference

Fly Models

Species	Genotype	PI/ Institution	Availability/Contact info	Publications
<i>D. melanogaster</i>	UAS- <i>PURA</i> ^Δ G179Wfs*22 ^Δ	Shinya Yamaoto, DVM., PhD	yamamoto@bcm.edu	-
<i>D. melanogaster</i>	UAS- <i>PURA</i> ^Δ A89P ^Δ	Shinya Yamaoto, DVM., PhD	yamamoto@bcm.edu	-
<i>D. melanogaster</i>	UAS- <i>PURA</i> ^Δ F 231C ^Δ	Shinya Yamaoto, DVM., PhD	yamamoto@bcm.edu	-
<i>D.</i>	UAS- <i>PURA</i> ^Δ F	Shinya	yamamoto@bcm.edu	-

<i>melanogaster</i>	233del [^]	Yamaoto, DVM., PhD	du	
<i>D. melanogaster</i>	UAS- <i>PURA</i> ^{^F} 271del [^]	Shinya Yamaoto, DVM., PhD	yamamoto@bcm.edu	-
<i>D. melanogaster</i>	<i>Pur-α</i> ^{^CR713} 82-KO-kG4 [^]	FlyBase	https://flybase.org/reports/FBaI0399808	

Antibodies

(*): Unknown

Antibody	Catalog #	Host species	Clonality	Species Reactivity	Applications	KO validated	Publications
Anti-PURA	ab79936	Rb	Polyclonal	Human, Mouse	WB, IF	*	(Gao et al., 2021)
Anti-PURA	ab125200	Rb	Polyclonal	Human	IP	*	(Gao et al., 2021)
PURA Antibody	3F10	Ms	Monoclonal	Human, Mouse, Rat	WB, ELISA, ICC/IF	*	(Kuo et al., 2020)