Prémio João Monjardino 2024 – Doenças Genéticas Raras: Deteção e Terapêutica

Resultados da avaliação

O Júri do Prémio João Monjardino 2024 reuniu-se no dia 24 de abril de 2025 para análise das candidaturas submetidas a concurso.

Após apreciação, o Júri deliberou, por unanimidade, atribuir o Prémio João Monjardino 2024 – *Doenças Genéticas Raras: Deteção e Terapêutica* a Jorge Diogo Cruz Ramos da Silva, pelo trabalho intitulado: *"Glucocorticoid receptor-dependent therapeutic efficacy of tauroursodeoxycholic acid in preclinical models of spinocerebellar ataxia type 3".*

O Júri decidiu igualmente não aceitar duas das candidaturas, por estas não preencherem o requisito relativo ao limite de idade do(a) candidato(a), conforme previsto no regulamento do concurso.

Nos termos do Código do Procedimento Administrativo, os(as) candidatos(as) dispõem de um prazo de **10 dias úteis** a contar da data deste aviso para, querendo, se pronunciarem em sede de audiência prévia, remetendo as suas observações para o endereço eletrónico: **premio.monjardino@fct.pt**.

Prémio João Monjardino 2024
Listagem de candidaturas submetidas a concurso

Nome completo	Título
Adriana Rocha Guimarães	*Role of MTHFR, IRF6, PAX7 and TP63 SNPs in susceptibility to non-syndromic orofacial cleft, a candidate gene study in a Portuguese population*
Ana Sofia Boasinha Ribeiro	*A human microglial cell model of autosomal recessive spastic ataxia of Charlevoix-Saguenay*
Artur Filipe Cardoso Duarte Rodrigues	*A Polymeric Nanoparticle Formulation for Targeted mRNA Delivery to Fibroblasts*
Carolina de Matos Pires	*Identification of novel candidate predisposing genes in familial nonmedullary thyroid carcinoma implicating DNA damage repair pathways*
Catarina Alexandra Catanas Madeira	*Functional and structural impact of 10 ACADM missense mutations on human medium chain acyl-Coa dehydrogenase*
Catarina da Cunha Ferreira	*Genotype-phenotype severity correlation in a multicentric Portuguese cohort of ABCA4-associated retinopathy*
Catarina Miranda Lourenço	*Impairment of adenosinergic system in Rett syndrome: Novel therapeutic target to boost BDNF signalling*

Cláudia Alexandra da Silva Rodrigues	*Personalized medicine: Function of CFTR variant p. Arg334Trp is rescued by currently available CFTR modulators*
Daniela Ferreira Melo	*Catalase, Glutathione Peroxidase, and Peroxiredoxin 2 in Erythrocyte Cytosol and Membrane in Hereditary Spherocytosis, Sickle Cell Disease, and? - Thalassemia*
Daniela Maria Grou Ramalho	*A non-genetic model of vascular shunts informs on the cellular mechanisms of formation and resolution of arteriovenous malformations*
Diogo Alexandre Costa Rodrigues	*Transthyretin monomers: a new plasma biomarker for pre-symptomatic transthyretin-related amyloidosis*
Diogo Miguel Barros Bitoque	*Molecular biology tools for the study and therapy of PDE6? mutations*
Elisabete Oliveira Teixeira	*Investigating USP42 Mutation as Underlying Cause of Familial Non-Medullary Thyroid Carcinoma*
Jorge Diogo Cruz Ramos da Silva	*Glucocorticoid receptor-dependent therapeutic efficacy of tauroursodeoxycholic acid in preclinical models of spinocerebellar ataxia type 3*
José García Peláez	*Genotype-first approach to identify associations between CDH1 germline variants and cancer phenotypes: a multicentre study by the European Reference Network on Genetic Tumour Risk Syndromes*
Lisbeth Selena Sousa e Silva	*Targeted next generation sequencing identifies novel pathogenic variants and provides molecular diagnoses in a cohort of pediatric and adult patients with unexplained mitochondrial dysfunction.*
Margarida Queirós da Silva Dias	*Mutational Spectrum and Deep Phenotyping in Pseudoxanthoma Elasticum: Findings from a Portuguese Cohort*
Maria Joana Valente Ribeiro	*Functional Recovery of a GCDH Variant Associated to Severe Deflavinylation-Molecular Insights into Potential Beneficial Effects of Riboflavin Supplementation in Glutaric Aciduria-Type I Patients*
Maria Miguel Rodrigues Gonçalves	*Portuguese Neonatal Screening Program: A Cohort Study of 18 Years Using MS/MS*
Marisa Ferreira Marques	*Ghrelin delays premature aging in Hutchinson-Gilford progeria syndrome*
Miguel Ângelo Jacinto	*Physical Activity, Exercise, and Sports in Individuals with Skeletal Dysplasia: What Is Known about Their Benefits?*
Pedro Emanuel Ferreira Peralta	*Is spastic ataxia 8 a protein misfolding disorder?*
Rebekah Koppenol	*The stress granule protein G3BP1 alleviates spinocerebellar*
Rute Ribeiro Pereira	*Characterization of CCDC103 profiles: further insights in primary ciliary dyskinesia and in human reproduction*

Sara Isabel Guerra Carvalhal Bandarra	*Biallelic BUB1 mutations cause microcephaly, developmental delay, and variable effects on cohesion and chromosome segregation*
Sara Neto Geada Batista	*Mutational Spectrum, Ocular and Olfactory Phenotypes of CNGB1-Related RP-Olfactory Dysfunction Syndrome in a Multiethnic Cohort*
Telmo José Martins Cortinhal	*Genetic profile of syndromic retinitis pigmentosa in Portugal*

FCT, 28 de maio de 2025