



Curriculum Vitae

Personal information **Cecile Rouzier**

Work experience

Physician in University Hospital

- start date 04/2011
- end date : present
- position held : praticien hospitalier
- employer's name : CHU de Nice
- country of employment : France

Education and training

- PhD : 2012 Université Côte d'Azur
- MD: 2008 Medical School of Nice

Additional information

Publications

Rouzier C, Pion E, Chaussonot A, Bris C, Ait-El-Mkadem S, Desquiret-Dumas V, Guegen N, Fragaki K, Amati-Bonneau P, Barcia G, Gaignard P, Steffann J, Pennisi A, Bonnefont JP, Lebogot E, Bannwarth S, Francou B, Rucheton B, Sternberg D, Martin-Negrier ML, Trimouille A, Hardy G, Allouche S, Acquaviva-Boursain C, Pagan C, Lèbre AS, Reynier P, Cossée M, Attarian S, Paquis-Flicklinger V, MITODIAG's network collaborators, Procaccio V. Primary mitochondrial disorders and mimics: Insights from a large French Cohort. ACTN, 2024, <https://doi.org/10.1002/acn3.52062>.

□ Mounir Serag, Morgane Plutino, Perrine Charles, Jean-Philippe Azulay, Annabelle Chaussonot, Véronique Paquis-Flucklinger, Samira Ait-El-Mkadem Saadi, **Cécile Rouzier**. A case report of *SYNE1* deficiency mimicking mitochondrial disease and the value of pangenomic investigations. 2023, Genes, accepté.

□ Ait-El-Mkadem Saadi S., Kaphan E., Jaurieta A., Fragaki K., Chaussonot A., Bannwarth S., Maues De Paula A., Paquis-Flucklinger V., **Rouzier C**. Splicing variants in *NARS2* are associated with milder phenotypes and intra-familial variability. Eur J Med Genet. 2022 Dec;65(12):104643. doi: 10.1016/j.ejmg.2022.104643. Epub 2022 Oct 14..

□ **Rouzier C**, Chaussonot A, Paquis-Flucklinger. Molecular diagnosis and genetic counseling for spinal muscular atrophy (SMA). V. Arch Pediatr. 2020 Dec;27(7S):7S9-7S14. doi: 10.1016/S0929-693X(20)30270-0. PMID: 33357600 Review.

□ Zereg E, Chaussonot A, Morel G, Bannwarth S, Sacconi S, Soriani MH, Attarian S, Cano A, Pouget J, Bellance R, Tranchant C, Lannes B, de Paula AM, Saadi Ait-El-Mkadem S, Chafino B, Berthet M, Fragaki K, Paquis-Flucklinger V, **Rouzier C**. Single-fiber studies for assigning pathogenicity of eight mitochondrial DNA variants associated with mitochondrial diseases. Hum Mutat . 2020 Aug;41(8):1394-1406. doi: 10.1002/humu.24037.

Projects

2025 Fondation Maladies Rares, Master's Grant in E-Health: *Functional classification of WFS1 gene variants through structural modelling and geometric learning*

2025 FILNEMUS Call for Proposals: *Implementation of a new technique for mitochondrial DNA quantification in single muscle fibres using digital PCR as a tool for interpreting variants of unknown significance in mitochondrial diseases* (15,000 euros)

2025 AOI, Nice University Hospital (CHU de Nice): *Implementation of a new technique for mitochondrial DNA quantification in single muscle fibres using digital PCR as a tool for interpreting variants of unknown significance in mitochondrial diseases* (15,000 euros)

2024 2024 Call for Proposals, Faculty of Medicine (UFR Médecine): *Combating diagnostic dead ends in rare diseases: transcriptomic approaches using RNA-seq and minigenes* (10,000 euros)

2023 AOI, Nice University Hospital (CHU de Nice): *Combating diagnostic dead ends in mitochondrial diseases:*

transcriptomic approaches on fibroblasts and blood cells (15,000 euros)

2023 FILNEMUS Call for Proposals: *Reducing diagnostic dead ends in mitochondrial diseases: transcriptomic approach* (12,000 euros)

2021 FILNEMUS Call for Proposals: *New tool for interpreting variants of unknown significance in single muscle fibres using NGS in mitochondrial diseases*

Memberships

Head of the Coordinating Reference Centre for Mitochondrial Diseases (CALISSON, certified in 2024, FILNEMUS network)

• **Coordinator of the CALISSON Multidisciplinary Team Meeting (RCP) for Adult and Paediatric Mitochondrial Diseases**, and of the **AURAGEN MITO-Nice RCP**

• **Coordination of the Reference Medical Biology Laboratories** for Mitochondrial Diseases, Wolfram Syndrome, and Spinal Amyotrophy within the Molecular Biology Unit of the Medical Genetics Department

• **Co-lead of the MITODIAG network since 2023**, together with Prof. Vincent Procaccio (11 laboratories involved in diagnosing mitochondrial diseases). Coordination of several working groups, including: harmonising the national diagnostic strategy, facilitating interaction with the PFMG2025 programme, and strengthening exchanges with research teams (cross-cutting working group with the FILNEMUS research commission)

• **Member of the Steering Committee of the Neuromuscular Disease Network (FILNEMUS) since 2022**

• **Co-coordinator, with Prof. Mireille Cossée, of the Molecular Diagnostic Commission of the Neuromuscular Disease Network (FILNEMUS) since 2024**

• **Co-lead of the NGS-DIAG network since 2017**, with Dr Jean Muller

(<https://ffgh.net/index.php/presentation/les-reseaux-partenaires/reseau-ngs-diagnostic>). Organisation of five scientific meetings since 2017, gathering more than 200 participants. Involvement in several working groups, including the national reference group on "Harmonising the interpretation of sequence variants generated by NGS analyses."

Member of the Board of Directors of the ANPGM (French National Association of Practitioners in Molecular Genetics) (2014–2022)

Other Relevant Information

Representative of the ERN-NMD within the "ESHG (European Society of Human Genetics) – ERN liaison group"

• **Member of the IT Commission of the Biology–Pathology Division**, involved in the implementation of the TD-GENET database

• **Member of the FILNEMUS Funding/Research Committee since 2023**

• **Member of the Board of Directors of the FFGH**; Member of the Scientific Committee of the *Assises de Génétique 2022*, Rennes (2021–2022)

• **Member of the National Committee for the Cross-Specialty Advanced Training (FST) in Clinical Molecular Genetics and Molecular Medicine** (2018–2022)

• **Member of the expert panel** evaluating practitioner accreditation applications for the *Agence de la biomédecine* (2017–2022)

• **Member of the National Qualification Commission in Medical Genetics** under the National Council of the Medical Association (since 2018)

• **Member of the Multidisciplinary Prenatal Diagnosis Centre of Nice University Hospital (CHU de Nice)** (member of the corum since 2015)