

# Leukodystrophies: novel perspectives on treatments and disease monitoring

## Paris, 25-27 September 2025

### Hotel Courtyard Paris Porte de Versailles

#### **Course description:**

Leukodystrophies are inherited white matter disorders of the brain. A first important step, especially in adult neurology, is the differential diagnosis with acquired forms of white matter disorders. The pathophysiology of leukodystrophies is complex and encompasses oligodendrocytopathies, astrocytopathies, microgliopathies, leuko-axonopathies and leuko-vasculopathies.

Novel biological and imaging markers are being developed to monitor disease onset, disease progression and response to treatment. These developments are especially important as there are increasing therapeutic options for leukodystrophies, from cell therapy (e.g., hematopoietic stem cell transplant) to gene therapy and small molecules. The acceleration of therapeutics has led to the development of new-born screening programmes for leukodystrophies, which also raise ethical questions.

#### **Learning objectives:**

- Introduction to leukodystrophies: definition, classification, MRI patterns, pathophysiology, main entities
- New molecular and imaging tools to diagnose and monitor disease progression in leukodystrophies
- Update on existing and emerging treatments in leukodystrophies (cell therapy, gene therapy, small molecules)
- Ethical challenges: burden of long-term MRI monitoring, impact of newborn screening, burden of therapeutic trials for children

#### **Target audience:**

Child neurologists, neurologists, neuroradiologists, paediatricians, geneticists, internists, neuroscientists

#### **Scientific Organising Committee:**

- Fanny Mochel – Reference centre for adult leukodystrophies, Pitié-Salpêtrière hospital, Paris, France
- Nicole Wolf – Amsterdam Leukodystrophy Centre, Amsterdam University medical centre, Amsterdam, the Netherlands
- Caroline Sevin – Reference centre for leukodystrophies, Bicêtre hospital, Bicêtre, France

# PROGRAM

## DAY 1

From 12 pm – arrival & registration & snack lunch available

1.15 – 1.30 pm: Welcome  
*Scientific Organising Committee*

### # Leukodystrophies – how to navigate?

**Chair : Fanny Mochel**

1.30 – 2.15 pm: **Leukodystrophies: definition, physiopathology**  
*Odile Boespflug-Tanguy*, Reference centre for leukodystrophies, Le Kremlin-Bicêtre hospital, Bicêtre, France

2.15-3.00 pm: **Classifications of leukodystrophies**  
*Odile Boespflug-Tanguy*, Reference centre for leukodystrophies, Le Kremlin-Bicêtre hospital, Bicêtre, France  
*Nicole Wolf*, Amsterdam Leukodystrophy Centre, Amsterdam University medical centre, Amsterdam, the Netherlands

3.00-3.30 pm: **Vascular leukoencephalopathies**  
*Dominique Herve*, Reference centre for rare neurovascular diseases, Lariboisière hospital, Paris, France

*3.30-4.00 pm: Coffee break*

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4.00-4.45 pm: **Leukodystrophies in adults**  
*Fanny Mochel*, Reference centre for adult leukodystrophies, Pitié-Salpêtrière hospital, Paris, France

4.45-5.15 pm: **Multimodal natural history studies in leukodystrophies for clinical trial readiness**  
*Adeline Vanderver*, Leukodystrophy Centre, Children's Hospital of Philadelphia, Philadelphia, USA

### # New tools to diagnose and monitor disease progression in leukodystrophies – part 1

**Chair: Caroline Sevin**

5.15-5.35 pm: **Quantified and standardized diffusion tensor imaging**  
*Fanny Mochel*, Reference centre for adult leukodystrophies, Pitié-Salpêtrière hospital, Paris, France

5.35-6.00 pm: **Perfusion MRI and application to adrenoleukodystrophy**  
*Patty Musolino*, Pediatric Stroke and Cerebrovascular Service, Massachusetts General Hospital, Boston, USA

7.30 pm **Buffet dinner at the hotel restaurant**

## DAY 2

### # New tools to diagnose and monitor disease progression in leukodystrophies – part 2

Chair: **Odile Boespflug-Tanguy**

#### 9.00-9.25 am: **NfL and plasma biomarkers in metabolic leukodystrophies**

*Johannes Berger*, Division of Pathobiology of the Nervous System, Medical University of Vienna, Vienna, Austria

#### 9.25-9.50 am: **Molecular signatures in Aicardi-Goutières syndrome**

*Yanick Crow*, Laboratory of Neurogenetics and Neuroinflammation, IMAGINE Institute, Paris, France

### # Therapies in leukodystrophies – part 1

Chair: **Odile Boespflug-Tanguy**

#### 9.50-10.40 am: **Hematopoietic stem cell transplantation in leukodystrophies – ALD, MLD, ALSP**

*Caroline Sevin*, Reference centre for leukodystrophies, Bicêtre hospital, Bicêtre, France

*Nicole Wolf*, Amsterdam Leukodystrophy Centre, Amsterdam University medical centre, Amsterdam, the Netherlands

*Fanny Mochel*, Reference centre for adult leukodystrophies, Pitié-Salpêtrière hospital, Paris, France

*10.40-11.00 am: Coffee break*

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11.00 am – 1 pm: **Workshops – 55 min parallel interactive sessions in small groups run 3 times in a row; participants attend each workshop once; 5 min for participants to switch between the 1st two sessions; lunch before the last session.**

#### Workshop 1: MRI-based recognition pattern

*Nicole Wolf*, Amsterdam Leukodystrophy Centre, Amsterdam University medical centre, Amsterdam, the Netherlands

*Odile Boespflug-Tanguy*, Reference centre for leukodystrophies, Le Kremlin-Bicêtre hospital, Bicêtre, France

#### Workshop 2: Hematopoietic stem cell transplantation in leukodystrophies

*Caroline Sevin*, Reference centre for leukodystrophies, Bicêtre hospital, Bicêtre, France

*Fanny Mochel*, Reference centre for adult leukodystrophies, Pitié-Salpêtrière hospital, Paris, France

#### Workshop 3: New-born screening in leukodystrophies

*Marc Engelen*, Pediatric neurology, Amsterdam University medical centre, Amsterdam, the Netherlands

*Alessandra Biffy*, Clinic and Laboratory of Pediatric Oncology and Stem Cell Transplantation, University of Padua, Padua, Italy

*1.00-2.00 pm: Buffet lunch*

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2.00-3.00 pm: **Workshops continued**

### # Therapies in leukodystrophies – part 2

Chair: **Nicole Wolf**

#### 3.00-4.00 pm: **Gene therapy in metabolic leukodystrophies – ALD, MLD, Krabbe, Canavan**

*Caroline Sevin*, Reference centre for leukodystrophies, Bicêtre hospital, Bicêtre, France

*Alessandra Biffy*, Clinic and Laboratory of Pediatric Oncology and Stem Cell Transplantation, University of Padua, Padua, Italy

*4.00-4.20 pm: Coffee break*

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4.20-5.20 pm: **Small molecules in leukodystrophies**

- **JAK inhibitors in interferonopathies**

*Yanick Crow*, Laboratory of Neurogenetics and Neuroinflammation, IMAGINE Institute, Paris, France

- **Leriglitazone in ALD**

*Patty Musolino*, Pediatric Stroke and Cerebrovascular Service, Massachusetts General Hospital, Boston, USA

*Fanny Mochel*, Reference centre for adult leukodystrophies, Pitié-Salpêtrière hospital, Paris, France

5.20-6.20 pm: **Gene therapies for hypomyelinating disorders**

- **TUBB4A, clinical trial readiness in H-ABC and ASO therapy**

*Adeline Vanderver*, Leukodystrophy Centre, Children's Hospital of Philadelphia, Philadelphia, USA

- **Gene therapy in PLP1**

*Odile Boespflug-Tanguy*, Reference centre for leukodystrophies, Le Kremlin-Bicêtre hospital, Bicêtre, France

7.15 pm [Departure for dinner](#)

7.45 pm [Dinner at the restaurant "L'île" \(Ile Saint Germain on the river Seine\)](#)

## DAY 3

**# Ethical challenges – part 1**

**Chair: Nicole Wolf**

9.00-9.30 am: **Impact of NBS in adult metabolic medicine – “toxic knowledge”**

*Marc Engelen*, Pediatric neurology, Amsterdam University medical centre, Amsterdam, the Netherlands

9.30-9.50 am: **Burden of long-term monitoring with MRI**

*Fanny Mochel*, Reference centre for adult leukodystrophies, Pitié-Salpêtrière hospital, Paris, France

9.50-10.20 am: **“Unethical trials”**

*Caroline Sevin*, Reference centre for leukodystrophies, Bicêtre hospital, Bicêtre, France

[10.20-10.50 am: Coffee break](#)

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**# Ethical challenges – part 2**

**Chair: Fanny Mochel**

10.50-11.20 am: **The need for academic registries, international guidelines, QoL outcome measures**

*Nicole Wolf*, Amsterdam Leukodystrophy Centre, Amsterdam University medical centre, Amsterdam, the Netherlands

11.20 am-12.15 pm: **The role of patient associations in the development of NBS programmes and access to (novel) therapies**

*Karen Harrisson*, Alex TLC, UK

12.15 – 12.30 pm: Wrap up & conclusion

*Scientific Organising Committee*

[12.30 pm: Snack lunch and departure](#)