



Interdisciplinary Workshop on Creatine Transporter Deficiency *SLC6A8*

PROGRAM

Monday, December 5th

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| 6:00 pm | Welcome |
| 6:45pm | Opening lecture : Intracerebral Creatine physiology
<i>Olivier Braissant, Lausanne, Switzerland</i> |
| 8:00 pm | Diner |

Tuesday, December 6th

Session 1: Natural History of Creatine Transporter Deficiency and relevant outcome

Chairmen: Brigitte Chabrol and Vincent des Portes

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| 8:00 am | Clinical data from a large European database
<i>Gajja S. Salomons, Amsterdam, The Netherlands</i> |
| 8:25 am | Clinical data from US patients
<i>Lumos Pharma, Austin, Texas, USA</i> |
| 8:50 am | Clinical data from a French cohort : the challenge of outcome measures
<i>Laurence Lion-François and Aurore Curie, Lyon, France</i> |
| 9:15 am | Patients' perspectives
<i>Xtraordinaire (France)</i>
<i>The Association for Creatine Deficiencies (USA)</i> |
| 9:45 am | Discussion panel |

Session 2 : Creatine transporter knockout mice

Chairmen: Hervé Moine and Olivier Braissant

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| 10:20 am | Ubiquitous and brain-specific Slc6a8 (exons 2-4) knockout mouse
<i>Matthew R. Skelton, USA</i> |
| 11:00 am | Ubiquitous Slc6a8 (exons 5-7) knockout mouse
<i>Laura Baroncelli, Pisa, Italy</i> |
| 11:35am | Discussion panel: strengths and weaknesses of Slc6a8 mouse models |
| 12:00 pm | Lunch |

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Session 3 : Therapeutics in Creatine Transporter Deficiency: towards new insights

Chairmen: Behrouz Kassai and Pascale de Lonlay

1:00 pm	Arginine <i>Roberta Battini, Pisa, Italy</i>
1:25 pm	Interest of S-adenosyl Methionine <i>Clara D.M. van Karnebeek, Vancouver, Canada</i>
1:50 pm	Cyclocreatine <i>Lumos Pharma, Austin, Texas, USA</i>
2:25 pm	Dodecyl creatine ester and lipid nanocapsule <i>Aloise Mabondzo, Paris, France</i>
3:00 pm	Preclinical data on novel creatine-derived compounds for creatine transporter deficiency <i>Maurizio Balestrino, Genova, Italy</i>
3:30 pm	Discussion panel

Session 4: Panel: Are we ready for new clinical trials?

Chairmen: Vincent des Portes, Olivier de Compiègne

4:00 pm	Pitfalls of placebo effect and outcome measures <i>Aurore Curie, Lyon, France</i>
4:25 pm	Regulatory challenges of therapeutics in rare disease <i>Fondation maladies rares, Christine Fétro, Lyon, France</i>
4:50 pm	Hopes and fears of patients and families <i>Xtraordinaire (France) and The Association for Creatine Deficiencies (USA)</i>
5:10 pm	Discussion panel

Scientific Board :

Pr Vincent des Portes, Filière DéfiScience,
Dr Aurore Curie, Filière DéfiScience
Dr Laurence Lion-François, Filière DéfiScience
Dr David Cheillan, Filière G2M
Pr Brigitte Chabrol, Filière G2M
Pr Pascale de Lonlay, Filière G2M
Madame Christine Fétro, Fondation Maladies Rares
Pr Behrouz Kassai, Réseau d'Investigation Pédiatrique des Produits de Santé
Messieurs Olivier de Compiègne et Franck de Franco, Association Xtraordinaire

Organizing committee :

Marie-Pierre Reymond, Filière DéfiScience
Victoria Déforges, Filière DéfiScience
Pr Vincent des Portes, Filière DéfiScience
Dr Aurore Curie, Filière DéfiScience