Programme

Thursday May 9

19.00-22.00 Welcome dinner at Comwell

Friday May 10

9.00 Welcome President, Peter Ott (Aarhus)

State of the art lectures
Chairs: Karl Heinz Weiss and Anil Dhawan

9.10 History of Wilson Disease James Dooley (London)
9.30 Diagnosis of WD in 2019 Aftab Ala (Guildford)
9.50 Classical and atypical neuro-psychiatric presentations Tomasz Litwin (Warsaw)
10.10 Specific challenges in pediatric Wilson patients Eve A. Roberts (Toronto)
10.30 Coffee break

Free papers
Chairs: Tomasz Litwin and Jan Pfeiffenberger

11.00 Major depressive disorder in patients with Wilson Disease and correlation with liver disease severity Michelle A. Camarata (Guildford)
11.15 Intestinal knockout of ATP7B impairs copper homeostasis Sarah Guttmann (Münster)
11.30 Wilson Disease: The French pediatric cohort Eduardo Couchonnal-Bedoya (Bron)
11.45 Evolution of cognitive and psycho behavioral profile after a liver transplant in case of neurological aggravation Carla Machado (Paris)
12.00 Cardiac tissue characterization in patients with Wilson Disease using cardiac magnetic resonance imaging Isabelle Mohr (Heidelberg)
12.15 Lunch
Friday May 10
Afternoon

Genotype-phenotype part I. Genetic and non-genetic factors
Chairs: Roman S. Polishchuk and Roderick Houwen

13.15 ATP7B-/- mice as an animal model for Wilson Disease
        Svetlana Lutsenko
        (Baltimore)

13.35 How to determine the functional consequences of a mutation using databases?
        Heinz Zoller (Innsbruck)

13.55 Nongenetic factors modifying the phenotype
        Albert Friedrich Stättermayer (Vienna)

14.15 Epigenetic factors in Wilson Disease
        Valentina Medici (Sacramento)

14.35 Is WD a mitochondrial disorder?
        Hans Zischka (Munich)

14.55 Coffee break and Poster session

Genotype-phenotype part II. How frequent is WD?
Chairs: Radan Bruha and Oliver Bandmann

15.55 Genetic and clinical prevalence in UK
        Oliver Bandmann (Sheffield)

16.10 Genetic and clinical prevalence in Sardinia
        Georgios Loudianos (Cagliari)

16.25 Genetic and clinical prevalence in France
        Quentin Beaulieu (Paris)

16.40 Analysis of the frequency and pathogenicity of ATP7B variants in a genomic dataset: Implications for Wilson Disease prevalence and penetrance
        Daniel Wallace (Brisbane)

Chairs: Peter Ferenci and Lisbeth Birk Møller

16.55 Clinical profile and management of Wilson Disease among children and adolescents in an Indian Tertiary Care Centre
        Seema Alam (New Delhi)

17.10 Genotype-phenotype correlation in India
        Harshad Devarbhavi (Bangalore)

17.25 Does clinical and genetic data fit?
        Peter Ott (Aarhus)

17.40 Panel discussion
        How frequent is WD.
        Is penetrance 100%?

17.55 End of day

19.00 Dinner at Ferdinand, Åboulevarden 28, 8000 Aarhus C
Saturday May 11
Morning

Studying the mutated protein
Chairs: Stephen G. Kaler and Sven C.D. van Ijzendoorn

9.00  Mutant interactome as a target for rescue of ATP7B function in Wilson Disease  Roman S. Polishchuk (Pozzuoli)

9.20  Structural studies by single-particle cryo-electron microscopy  Poul Nissen (Aarhus)

9.40  Copper binding to human P-type ATPase ATP7B  Nina Salustros (Copenhagen)

9.55  Searching for a new treatment of Wilson Disease  Christine J. F. Nielsen (Aarhus)

10.10  Differentiation of pluripotent stem cells to bile canalliculi-forming hepatocytes to study genetic liver diseases involving hepatocyte polarity  Arend W. Overeem (Groningen)

10.25  Coffee break

Assessing copper metabolism
Chairs: Svetlana Lutsenko and Valentina Medici

11.00  Quantifying changes in the KF-Ring  Jesper Hjortdal (Aarhus)

11.15  In exchangeable copper we trust  Nouzha Djebirani-Oussedik (Paris)

11.30  Calculated free copper is better  Frederick K. Askari (Ann Arbor)

11.45  Evaluation of human Copper metabolism and the diagnosis of Wilson Disease by $^{64}$CuCl$_2$ positron emission tomography  Thomas Damgaard Sandahl (Aarhus)

12.00  Substantia nigra hyperechogenicity change over time in patients with Wilson Disease  Marta Skowronska (Warsaw)

12.15  CRISPR/CAS9-mediated correction of mutated copper transporter ATP7B  Vanessa Sandfort (Münster)

12.30  Lunch
**Saturday May 11**
**Afternoon**

**Workshop Session 1:**  
**Recommendations for clinical trials in WD**  
*Chairs: Aurélia Poujois and Peter Ott*  
*Moderators: Eve A. Roberts and Peter Ferenci*

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<tr>
<td>13.30</td>
<td>1</td>
<td>Potential study designs in orphan disease</td>
<td>Ralf-Dieter Hilgers</td>
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<td>Moderator Comments and questions</td>
<td>Peter Ferenci (Vienna)</td>
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<td>14.30</td>
<td>2</td>
<td>Study Endpoints – Liver</td>
<td>Michael L. Schilsky</td>
<td>New Haven</td>
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<td>Moderator Comments and questions</td>
<td>Eve A. Roberts (Toronto)</td>
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**Workshop Session 2:**  
*Chairs: Frederick K. Askari and Aftab Ala*  
*Moderators: Michael L. Schilsky and Karl Heinz Weiss*

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<tr>
<td>16.00</td>
<td>3</td>
<td>Study Endpoints – Neurology and quantifying neurological changes</td>
<td>Anna Czlonkowska</td>
<td>Warsaw</td>
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<td>16.20</td>
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<td>Moderator Comments and questions</td>
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<td>Monitoring the patients</td>
<td>Peter Ferenci (Vienna)</td>
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<td>Moderator Comments and questions</td>
<td>Karl Heinz Weiss</td>
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<td>18.30</td>
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<td>Dinner at NO16, Europaplads 16, 8000 Aarhus C</td>
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Sunday May 12
Morning

**Treatment**  
*Chairs: Eve A. Roberts and Piotr Socha*

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<tr>
<td>9.00</td>
<td>Treatment of Wilson Disease</td>
<td>Peter Ferenci (Vienna)</td>
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<td>9.30</td>
<td>Liver transplantation in adults with WD</td>
<td>Rodolphe Sobesky (Villejuif)</td>
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<tr>
<td>9.45</td>
<td>Liver transplantation in children with WD</td>
<td>Anil Dhawan (London)</td>
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<td>10.00</td>
<td>Controversy: Liver transplantation in neurologic disease</td>
<td>Aurélie Poujois (Paris)</td>
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<td>10.15</td>
<td>Brain atrophy and neurological impairment in Wilson Disease</td>
<td>Lukasz Smolinski (Warsaw)</td>
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<td>Coffee break</td>
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**Future treatments**  
*Chairs: Anna Czlonkowska and Petr Dusek*

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<tr>
<td>11.00</td>
<td>Medical needs not addressed by current treatments</td>
<td>Piotr Socha (Warsaw)</td>
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<td>11.15</td>
<td>Viral gene therapy for Menkes disease and related Copper Transport Disorders</td>
<td>Stephen G. Kaler (Bethesda)</td>
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<td>11.30</td>
<td>Emerging therapies: hepatocyte transfer</td>
<td>Sanjeev Gupta (Bronx)</td>
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<td>11.45</td>
<td>Evidence of Erad on ATP7B mutations indicates pharmacological chaperones as an optional individualized therapy for Wilson Disease</td>
<td>Christina Hund (Rostock)</td>
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<td>12.00</td>
<td>Gene therapy</td>
<td>Gloria González-Aseguinolaza (Pamplona)</td>
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<td>12.15</td>
<td>Closing remarks</td>
<td>Peter Ott (Aarhus)</td>
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<td>12.25</td>
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<td>13.30</td>
<td>Departure</td>
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