

PROGRAM

Cerebral Folate Deficiency (CFD)

1st International Meeting

Friday 17th and Saturday 18th of May 2019
Lecture Hall, University Childrens' Hospital Zurich



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*Das Spital der
Eleonorenstiftung*

Timely therapy makes a difference!



Ten years ago, cerebral folate transport deficiency has been described to be caused by loss of function mutations in the *FOLR1* gene coding for folate receptor alpha. Since then numerous reports have confirmed and delineated this inherited disorder of folate transport into the brain. The lack of biological markers outside the brain is a unique feature of this disease that differentiate it from other folate metabolic disorders such as hereditary folate malabsorption or methyl tetrahydrofolate reductase deficiency. It is certainly one of the reasons for the

delay in diagnosis and thus treatment for cerebral folate deficiency caused by *FOLR1* mutation or by any other gene defect related to folate metabolism.

The purpose of this 1st international meeting on cerebral folate deficiency disorders is to enhance the public awareness for this group of diseases and to advocate for the need of early diagnosis to prevent the occurrence of irreversible neurological symptoms. The past ten years have demonstrated that for many disorders of folate metabolism and transport patient outcomes correlates with the begin of treatment. In respect of the *FOLR1* gene defect the preventive therapy before the onset of neurological symptoms goes along with a complete normal psychomotor development. Thus, the timely therapy determines the fate of the affected children and their families and therefore this must be aimed for.

I would like to express my gratefulness to colleagues, families and friends that helped to organize this conference and that will certainly contribute to and enrich this meeting.

Robert Steinfeld, MD, PhD
Head of Department of Pediatric Neurology
University Childrens' Hospital Zurich

**1st international Meeting on
Cerebral Folate Deficiency (CFD) Disorders
17th & 18th May 2019
Lecture Hall, University Childrens' Hospital Zurich**

Program

Friday, 17th May 2019

Current diagnosis and therapy of CFD Disorders

08:00 Registration, Coffee & Tea

09:00 Opening Robert Steinfeld – Zürich

Biochemistry of Folates

09:15 **Folate Metabolism** Sean Froese – Zurich

09:45 **Transcriptional Regulation of Proteins
Involved in Folate Metabolism** Richard Finnell – Houston

10:15 Coffee Break

Folate Pathway and Transport defects

10:45 **Hereditary folate malabsorption** David Goldman – New York

11:15 **Cerebral folate transport deficiency** Robert Steinfeld – Zurich

11:45 **Folate Metabolic Disorders and
Secondary Causes of CFD** Shamima Rahman – London

12:15 Lunch & Networking

Monitoring and Treatment

14:00 **Brain Imaging of CFD** Steffi Dreha-Kolaczewski – Göttingen

14:30 **10 Years treating CFD** Gabriela & Robert Bopp – Ellwangen,
Robert Steinfeld – Zurich

15:30 Coffee Break

Clinical Spectrum of CFD Disorders

16:00 **Free Communications** (15' each)

17:30 **Ask the Expert – Questions from patient families and colleagues**
S. Dreha-Kolaczewski, R. Finnell, D. Goldman, S. Rahman, R. Steinfeld

Saturday, 18th May 2019

Guidelines for diagnosis and therapy of CFD Disorders

08:00 Registration, Coffee & Tea

09:00 Newborn Screening

Ralph Fingerhut – Zurich

09:30 **Consensus on diagnosis and treatment of CFD disorders**

- Newborn screening
- Diagnostic criteria
- Folate treatment
- Other therapies
- Therapy Monitoring
- Long Term Follow up (into adulthood)
- Patient registry

The CFD Foundation presents itself

- Establishment of corresponding organizations in other countries
- International Website
- Blogging platform
- Newborn Screening

11:00 Coffee Break

11:30 Presentation of Consensus Results

12:30 Lunch Sandwiches and Farewell

Registration until 30th April

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Venue

Lecture Hall (Hörsaal)
University Children's
Hospital Zurich
Steinwiesstrasse 75
8032 Zürich

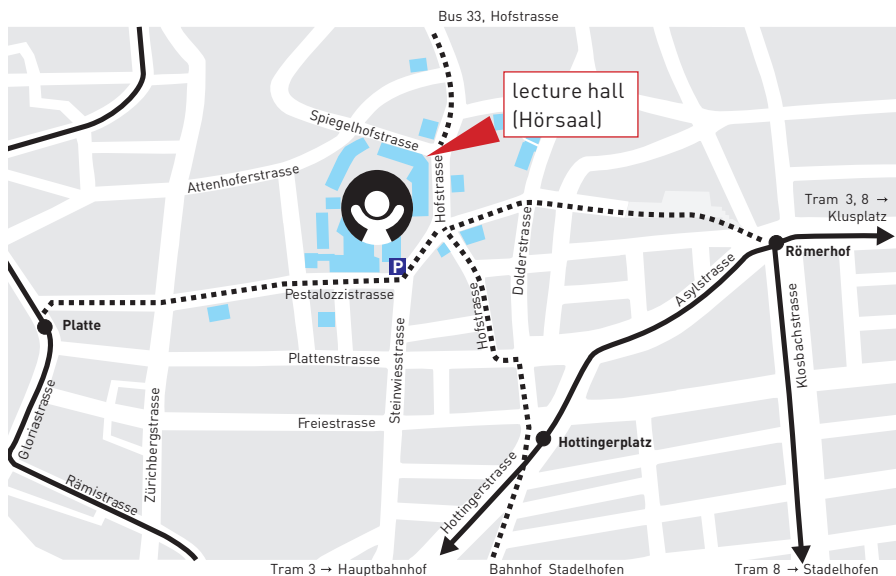


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How to find us

From the main railway station

- Tram 3 Towards Klusplatz to Hottingerplatz stop*
- Tram 6 Towards Zoo to Platte stop*

From Stadelhofen

- Tram 8 Towards Klusplatz to Römerhof stop*

* The Children's hospital is approximately a 10-minute walk from these stops.