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Strasbourg, June 7th 2010

Dear Colleagues,

You have been informed by Jean-Pierre FRYNS that the "**Twenty-first European Meeting on Dysmorphology**" will be in Strasbourg on **September 2 and 3, 2010**.

The meeting and housing will be in "**Le Bischenberg**" which is a nice meeting place located in the Vosges mountains, 20km West from Strasbourg.

The meeting will start on **Thursday, September 2 at 8.00am.**, and it will end on **Friday, September 3 at 11pm**. *Arrival on Wednesday September 1, late afternoon.*

Please could you return to me before **JULY 6**, the enclosed registration form and abstract form (a copy of the abstract form (see guidelines) has to be sent to J.P. FRYNS).

Please, could you send these forms preferentially by e-mail ?

Remember that it was decided that only **one presentation per participant** will be possible.

Thanking you in advance.

Yours sincerely

Pr Claude STOLL

P.S. The 4 sessions will be on:

1. **MCA/MR syndromes**
2. **Fetal pathology**
3. **New chromosomal microdeletion syndromes**
4. **Connective tissue disorders**

There will also be a session on "**Unknown**".

21st EUROPEAN MEETING ON DYSMORPHOLOGY
Strasbourg, France, September 02-03 2010

FULL NAME

ADDRESS

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ABSTRACT FORM

(Title, Authors, Affiliations, Text).

To be sent to :
Pr. C. STOLL
Laboratoire de Génétique Médicale
Faculté de Médecine
11,rue Humann
67085 STRASBOURG Cédex (France)
Fax(33)3.68.85.31.79
E-mail:Claude.Stoll@medecine.u-strasbg.fr

Please, send a copy to :
Pr. Dr. J.P. FRYNS
Center for Human Genetics
U.Z.Gasthuisberg
Herestraat,49
B-3000 LEUVEN(Belgium)
Fax (+32)16.34.60.51
E-mail: Jean-Pierre.Fryns@med.kuleuven.be

Not later than July 6, 2010

21st EUROPEAN MEETING ON DYSMORPHOLOGY

Strasbourg, France, September 02-03, 2010

TO BE RETURNED NOT LATER THAN JULY 6, 2010

REGISTRATION FORM :

NAME FIRST NAME

STREET.....

CITY.....

COUNTRYPHONE.....FAX.....

E-MAIL.....

AFFILIATION

REGISTRATION FEES

530 € (five hundred thirty Euros)

This fee includes documentation, the meals and the hotel accommodation : September 01, 02 and 03.

Total amount due has to be sent **before July 6, 2010** to :

Banque Populaire d'Alsace, Agence Ancienne Douane

Code 17607-00001 - Acct : " APPROMERE ", N° 06193794351 Rib 91

IBAN:FR76 1760 7000 0106 1937 9435 191 -BIC: CCBPFRPPSTR

Please, don't forget to give your name when sending the money and be sure that **530 € will really be paid to our bank** and specify "**FREE OF CHARGE FOR APPROMERE**". Any charges for banking fees or incorrect remittance of registration fees will be collected on site.

ARRIVAL DATE.....

By (car [], plane [], train [])

From.....To.....Arrival time.....

DEPARTURE DATE.....

RETURN BEFORE JULY 6, 2010 to :

Pr. Claude STOLL

Laboratoire de Génétique Médicale

Faculté de Médecine, 11, rue Humann

67085 STRASBOURG Cédex, France

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E-mail: Claude.Stoll@medecine.u-strasbg.fr

Abstract Guidelines

PLEASE PREPARE YOUR ABSTRACT ACCORDING TO THE FOLLOWING EXAMPLE:

SEVERE HYPERNATREMIC DEHYDRATION IN AN INFANT WITH NETHERTON SYNDROME

Y. ALEMBIK¹, D. TCHOMAKOV², E. HEID³, N. BOEHM, J.MESSER² and C.STOLL¹

¹ Service de Génétique Médicale

² Service de Pédiatrie

³ Service de Dermatologie, Centre Hospitalo-Universitaire, Strasbourg, France.

Autosomal recessive congenital ichthyosis (ARCI) is a group of inherited disease of cornification in which progress has recently been made in the identification of pathogenic mechanisms causing the disorder. Transglutaminase 1 (TGM 1) has been found as a defective gene in a large fraction of patients with lamellar ichthyosis. More recently the mutation of SPINK 5 was described in the Netherton syndrome. Netherton syndrome is a rare ARCI characterized by ichthyosis and the characteristic hair abnormality trichorrhexis invaginata. We report a patient with the severe hypernatremic dehydration form of the Netherton syndrome.

Infant G. was the first child of consanguineous parents. Ichthyosis was present at birth. He was admitted to intensive care at the age of 4 days for an important loss of weight and dehydration. Severe hypernatremia and convulsions occurred. Despite intensive care the baby died at the age of 11 days. The diagnostic of Netherton syndrome was confirmed by the pathognomonic hair shaft anomaly, trichorrhexis invaginata (bamboo hair). Skin biopsy showed premature lamellar body secretion and foci of electron-dense material in the intercellular spaces of stratum corneum which are relatively specific markers for Netherton syndrome. These abnormalities could explain the impaired permeability barrier in Netherton syndrome, and account for hypernatremia and dehydration in infants with the syndrome. Netherton syndrome is characterized by a large variability of phenotype expressivity. The major neonatal complication is the hypernatremic dehydration, which can be fatal as in this patient or complicated by neurologic signs (intracranial hemorrhage) and secondary sequellae.

Title: Capitals/boldface

Authors: Initials + surname in capitals/italics

Affiliations: numbered, superscript (see example)

Please send a copy of your abstract preferentially by e-mail or on floppy disk to:

Prof. Dr. J.P. FRYNS

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