Registries for patients with undiagnosed rare diseases

The correct and timely diagnosis of rare diseases is a common challenge. Therefore, the German Action Plan on Rare Diseases recommends the development of a national registry for patients with undiagnosed rare diseases. In this context, a distinction will be made between patients with a tentative diagnosis and patients with an as yet unknown rare disease.

Objectives of the workshop

It is the overall objective of the workshop to analyse, whether it is feasible and helpful to collect information on patients with undiagnosed diseases within a registry, in order to enable or accelerate diagnosis and/or adequate therapy. In particular, the following questions shall be addressed:

- What are the options to collect information on patients with an undiagnosed rare disease as a basis for future research and diagnostic evaluation?
- What are the key issues/challenges of national registries for undiagnosed rare diseases?
- How should such registries be integrated into European/international initiatives?
- How could a registry for undiagnosed rare diseases be linked efficiently to interested researchers in the field?

The workshop will be held in English, in order to enable international guests to present their projects and participate in the discussion.

Organizers

TMF - Infrastructures for Medical Research

The TMF is the umbrella organization for networked medical research in Germany. It is the platform for interdisciplinary exchange as well as cross-project and cross-location cooperation in order to identify and solve the organizational, legal/ethical and technological problems of modern medical research. Solutions range from expert opinions, generic concepts, and IT applications to checklists, practical guides, training, and consultation services. The TMF makes these solutions available to the public free of charge.

www.tmf-ev.de

Research for Rare

Since 2003, the German Federal Ministry of Education and Research has been sponsoring research networks in their endeavours to try to understand the causes of rare diseases and to develop urgently needed new therapeutic approaches. Networking and the confederation of specialists on specific disease groups can counteract the problem of small patient numbers and enables the concentrated collection and evaluation of information. To foster closer collaboration of the research networks on rare diseases, a board of speakers as decision-making and coordinating body for the overarching collaboration was called into existence. Their office serves as liaison between the research networks, facilitates the exchange of information and supports the board of speakers in the implementation of its projects.

www.research4rare.de

In co-operation with

NAMSE
Nationales Aktionsbündnis für Menschen mit Seltenen Erkrankungen

Funded by

Federal Ministry of Education and Research

TMF-Workshop

Registries for patients with undiagnosed rare diseases

21 November 2013 | Berlin

RESEARCH FOR RARE
Forschung für seltene Erkrankungen

TMF

Program

Version: 13.08.2013

10.30 Welcome and introduction
Cornelia Zeidler (Hannover Medical School)
Miriam Schlangen (External Coordination Office, National Action League for People with Rare Diseases)

10.40 The Undiagnosed Diseases Program of the NIH
Cynthia Tifft (National Institutes of Health)

11.00 The European Plattform for Rare Diseases
Jaroslaw Waligora (European Commission)

11.20 The Undiagnosed Diseases Program of the Netherlands
Wendy van Zelst-Stams (University Medical Center St Radboud, Nijmegen)

11.40 Patients with Undiagnosed Rare Diseases - The EUCERD Perspective
Ségolène Aymé (French Institute of Health and Medical Research, INSERM)

12.00 Panel Discussion
Cynthia Tifft, Jaroslaw Waligora, Ségolène Aymé, Wendy van Zelst-Stams, N.N.

12.45 Summary / key issues
Thomas O. F. Wagner (University Hospital, Frankfurt/M)

13.00 Lunch

14.00 Compilation of phenotypes HPO/Phenomizer
Peter Robinson (Institute for Medical Genetics, Charité)

14.20 Decipher
Jawahar Swaminathan (DECIPHER – Wellcome Trust Sanger Institute)

14.40 Study of Health in Pomerania (SHIP)
Carsten-Oliver Schmidt (University of Greifswald)

15.00 Panel Discussion: What do we need for a successful registry?
Cynthia Tifft, Jaroslaw Waligora, Ségolène Aymé, Wendy van Zelst-Stams, Peter Robinson, Helen Firth *(tbc)*, N.N.

15.45 Coffee Break

16.00 Resume of Results / Next Steps

16.45 Closing Remarks
Cornelia Zeidler

Evening: Get together

Organisational Issues

Date:
21 November 2013, 10.30- 17.00

Venue:
TMF conference room
Charlottenstraße 42/Dorotheenstraße
10117 Berlin | Germany
Travel information and location map included

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Registration:
Participation is free of charge. Please register on-line by 14 November 2013 at www.tmf-ev.de/anmelden.

Scientific Organisation:
- Christoph Klein, Munich
- Christine Mundlos, German Alliance for Rare Diseases (ACHSE), Berlin
- Christiane Steinmüller, Federal Ministry of Education and Research, Bonn
- Frank Ückert, Mainz
- TOF Wagner, Frankfurt
- Maggie Walter, Munich
- Cornelia Zeidler, Hannover