



RESEARCH **FOR** RARE
Forschung für seltene Erkrankungen



Bundesministerium
für Bildung
und Forschung

Workshop Registries for patients with undiagnosed rare diseases

Workshop

Registries for patients with undiagnosed rare diseases

For some patients with rare diseases a complete diagnosis is not possible even with the highest levels of medical knowledge.

Programs are required

- to reduce the uncertainties for the patients with an unclear diagnosis
- to provide the best possible chance of achieving a precise diagnosis and
- to allow access to appropriate care once a diagnosis is achieved.



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Workshop

Registries for patients with undiagnosed rare diseases

Discussion about patients with unclear diagnosis

- How can we increase the chance for a precise diagnosis?
- How many patients exist in Germany?
- How can we improve access for researchers and clinical trials?

Background

Can a registry for patients with undiagnosed or better unclear rare diseases help to achieve a precise diagnosis?

Discussion in two separate working groups on registries

1) Research prospective

WG was established by the German networks on rare diseases funded by the German Ministry for Research and Education (BMBF)

2) National Action Plan (NAMSE)



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Workshop

Registries for patients with undiagnosed rare diseases

Key questions

Is a registry for patients with unclear/undiagnosed rare diseases a promising and feasible approach to increase the chance for a precise diagnosis?

What are the requirements for a successful program?



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