FACT SHEET

Solve RD
Solving the Unsolved Rare Diseases

The Solve-RD cohorts

Solve-RD aims to find a diagnosis for rare disease patients who did not get a molecular diagnosis so far. We will compare the potential of different -omics technologies regarding their contribution to solve the pathogenicity in different patient cohorts, reaching from neurodevelopmental to late onset diseases including diagnostic specialties like cancer. The clinical expertise of numerous European Reference Networks (ERNs) is integrated by clinically well selected patient cohorts. This FACT SHEET gives an overview of the four Solve-RD cohorts of rare disease patients which have been defined.

1 UNSOLVED CASES*

Definition: unsolved RD case with an inconclusive exome/genome

Number: at least 19.000 unsolved exomes/genomes

Main activities: standardized collation and re-analysis

**in collaboration with all ERNs, Undiagnosed Disease Initiatives and further associated partners*

2 SPECIFIC ERN COHORTS

Definition: Specific cohorts from four core ERNs

Number: a) 2.000 WGS to achieve a more complete (non-)coding sequence & Svs etc.; b) 500 long-read WGS; c) >2.000 cases novel omics approaches

Main activities: „beyond the exome“ approaches

3 ULTRA RARE RARE DISEAES

Definition: phenotypically most special/remarkable patients with a rare disease without an exome

Number: 800

Main activities: Phenotype jamborees and exome analysis

4 THE UNSOLVABLES

Definition: Highly recognizable clinically defined diseases/ syndromes for which no disease gene was identified yet despite WES/WGS and considerable research invested

Number: 120

Main activities: All -omics tools to ‚crack' the „Unsolvables“

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