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: Rare disease cases with an inconclusive exome/genome

Number: 19,000 unsolved exomes/genomes

Main activities: Perform standardised collation and re-analysis

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

2 SPECIFIC ERN COHORTS

Definition: Disease group specific cohorts from four core ERNs (exome available)

Number: a) 2,000 WGS for more complete (non-)coding sequence & CNV/SVs etc.;

b) 500 long-read WGS;

c) >2,000 cases novel omics approaches

Main activities: Conduct „beyond the exome“ approaches

3 ULTRA RARE RARE DISEASES

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

Number: 1,200 exomes (300 per core ERN)

Main activities: Carry out phenotype jamborees and exome analysis

4 THE UNSOLVABLES

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

Number: 120 syndromes/ diseases

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

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