Solving the unsolved Rare Diseases

Informationsblatt

Projektinformationen

Solve-RD
ID Finanzhilfevereinbarung: 779257

Projektwebsite

Startdatum 1 Januar 2018
Enddatum 31 Dezember 2022

Finanziert unter
H2020-EU.3.1.1.

Gesamtbudget € 15 361 621,25

EU-Beitrag € 15 361 621,25

Koordiniert durch
EBERHARD KARLS
UNIVERSITAET TUEBINGEN
Deutschland

Ziel

The main ambitions of the Solve-RD proposal are (i) to solve large numbers of RD, for which a molecular cause is not known yet, by sophisticated combined Omics approaches, and (ii) to improve diagnostics of RD patients through a “genetic knowledge web”. Solve-RD will pursue a clear visionary and integrated “beyond the exome” approach. The entire Solve-RD proposal has been motivated, designed and put together by a core group of four ERNs, but also reaches out to all 24 ERNs. To tackle diseases which are unsolved by applying cutting edge strategies, Solve-RD has thus formed a consortium that comprises (i) leading clinicians, geneticists and translational researchers of these ERNs, (ii) RD research and diagnostic infrastructures, (iii) patient organisations, as well as (iv) leading experts in the field of -omics technologies, bioinformatics and knowledge management. Solve-RD will deliver 7 main implementation steps: (i) Collect Phenotypes, (ii) New phenotype
patterns, (iii) Re-analyse exomes / genomes, (iv) Novel molecular strategies, (v) Functional analysis, (iv) Clinical utility and (vii) Towards therapy. For analysis Solve-RD will apply data driven and expert driven approaches. We anticipate to increase diagnostic yield from 19,000 unsolved exomes/genomes by about 3-5%. Cohort specific innovative -omis strategies will be pursued, also addressing cost-effective issues. Analysis of more than 800 patients with highly peculiar (ultra-rare) phenotypes will highly increase the chance to find novel disease genes and novel disease mechanisms. We anticipate to solve more than 2,000 cases. Finding further matching patients will be secured by newly developed matchmaking approaches and by screening using MIPs technology in the more than 20,000 unclassified patients of the ERNs. For the first time in Europe we will also implement a novel brokerage structure connecting clinicians, gene discoverer and basic researcher to quickly verify novel genes and disease mechanisms.

Wissenschaftliches Gebiet

Programm/Programme

Thema/Themen

Aufforderung zur Vorschlagseinreichung

H2020-SC1-2017-Single-Stage-RTD

Finanzierungsplan

RIA - Research and Innovation action

Koordinator

EBERHARD KARLS UNIVERSITAET TUEBINGEN

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Aktivitätstyp
Higher or Secondary Education Establishments

EU-Beitrag
€ 2 718 962,50

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