

Univerza
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Diagnosis and treatment of rare disorder Hereditary Erythrocytosis

Research & Innovation Transforming European Healthcare
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Prof. dr. Nataša Debeljak

Medical Centre for Molecular Biology, Institute of Biochemistry and Molecular Genetics
Faculty of Medicine, University of Ljubljana, Slovenia

natasa.debeljak@mf.uni-lj.si
<https://www.mf.uni-lj.si/ibk/>

Short history of the Institute

Univerza
v Ljubljani

Medicinska
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University of Ljubljana
Faculty of Medicine
*Institute of Biochemistry
and Molecular Genetics*

- Faculty of Medicine established 1919 as one of the founding faculties of the University of Ljubljana
<https://www.mf.uni-lj.si/>
- the oldest higher education teaching and scientific research institution in the fields of medicine, dental medicine and the biomedical sciences in Slovenia
- Institute of Biochemistry and Molecular Genetics one of the bigger preclinical institute at Faculty of Medicine
<https://www.mf.uni-lj.si/ibk/>
- 5 laboratories and 2 centre
- Mission: education and research in the field of biochemistry and molecular biology, both in basic research, as well as in translational research and the transfer of new technologies into clinical practice
- Medical Center for Molecular Biology is focusing on field of medical molecular biology and genetics

Facts on rare disorders

- more than 6000 distinct rare diseases in the EU
- each one affects fewer than 1 in 2000 people
- altogether up to 36 million people in the EU live with a rare disease
- around 80% of rare diseases are of genetic origin
- onset of 70% of rare diseases is in childhood
- no cure for the vast majority of rare diseases

Hereditary Erythrocytosis



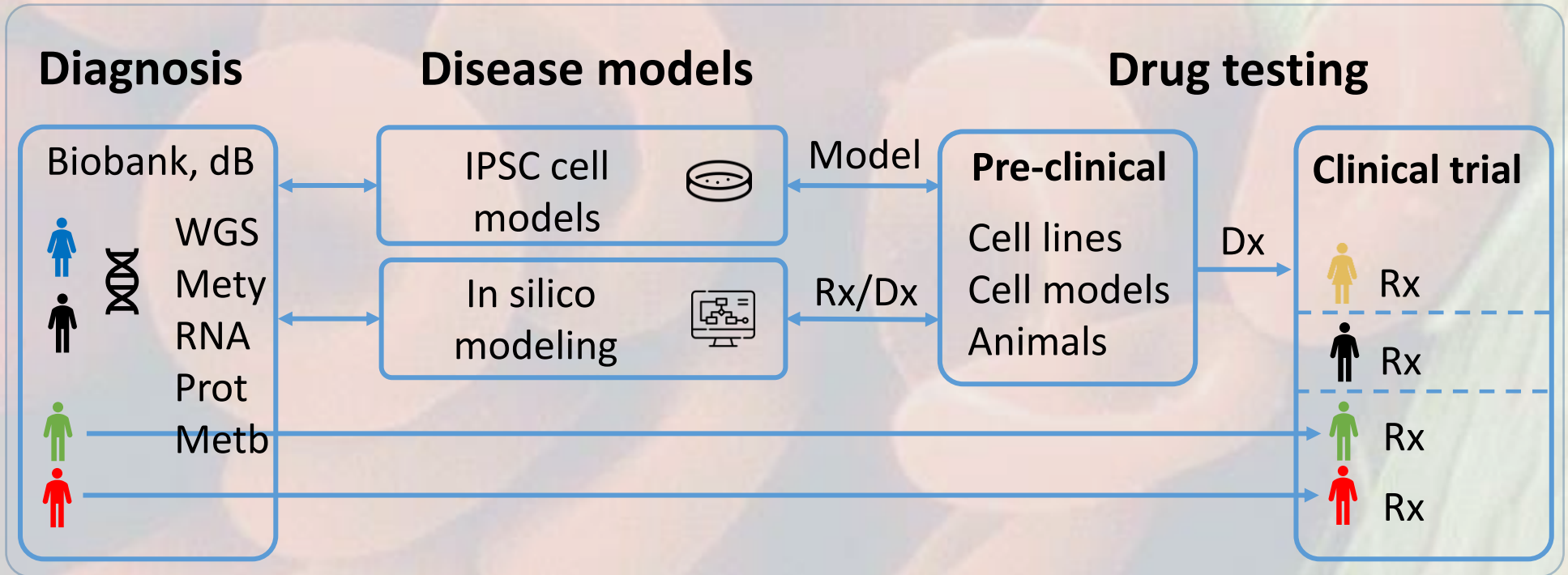
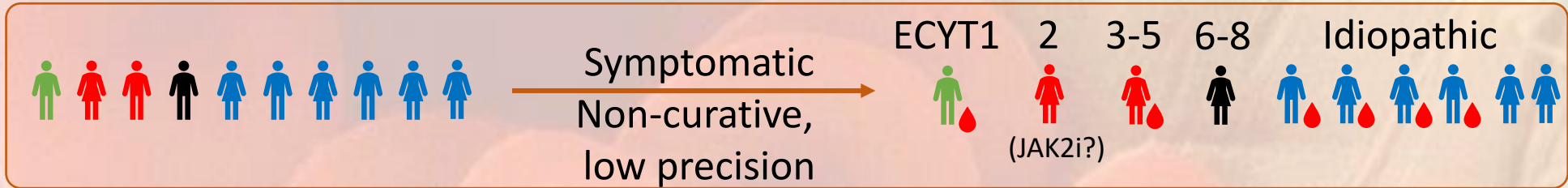
8 subtypes of rare disorder
affecting >2000 people
thromboembolic complications

4-9 years to diagnosis
>8 visits to doctors
>4 different specialists

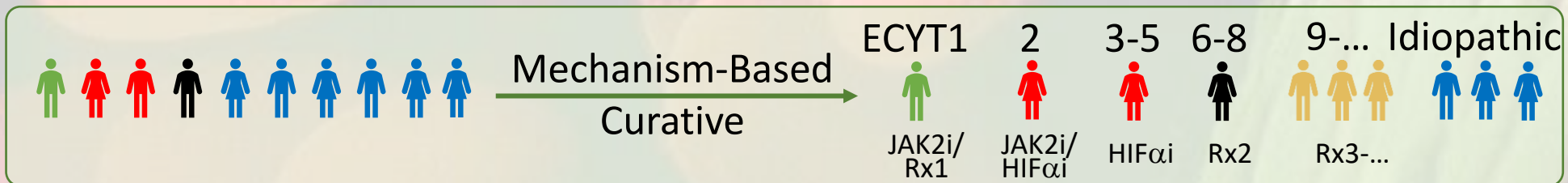
9 known causative genes
>60% without diagnosis
symptomatic treatment



Current Diagnosis and Treatment



Future Diagnosis and Treatment



Open to collaboration in the field of:

Diagnosis



Drug testing



Disease models

Contact:

Prof. dr. Nataša Debeljak

natasa.debeljak@mf.uni-lj.si

<https://orcid.org/0000-0003-1294-292X>