Expert Conference on Rare Diseases

Session II Early Diagnosis for Rare Diseases

Panel

Early diagnosis: from newborn screening to personalized patient care

Moderator

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ISNS International Society for Neonatal Screening

European Reference Network for rare or low prevalence complex diseases

Network Hereditary Metabolic Disorders (MetabERN)

Member General University Hospital in Prague — Czechia

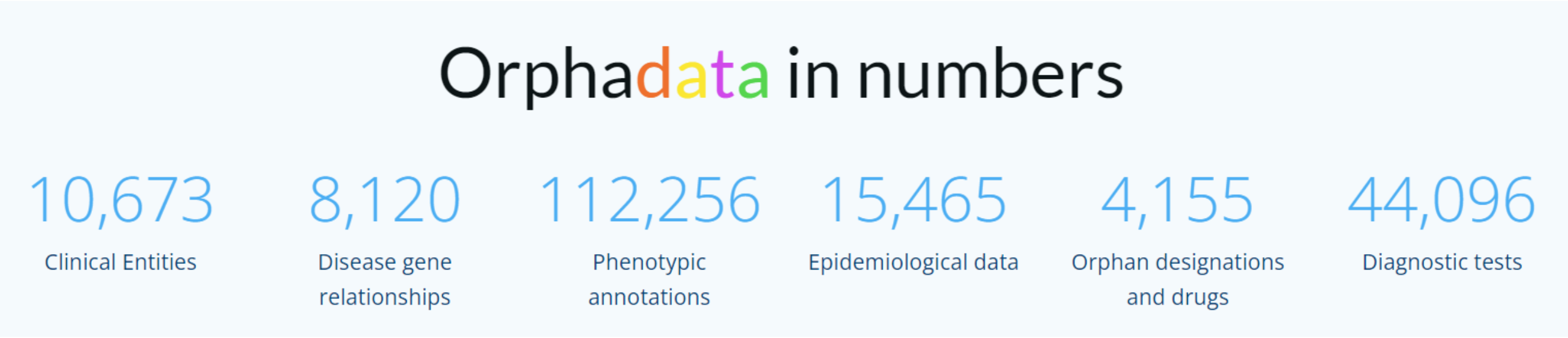
Program overview

17:15- 17:35 PRESENTATIONS

- Diagnostic pathways for RD
- IVDR 2017/746
- Key indicators of newborn screening
- Newborn dried blood spot screening and follow up in Sweden

17:35-18:00 PANEL DISCUSSION

Orphadata as of October 20, 2022



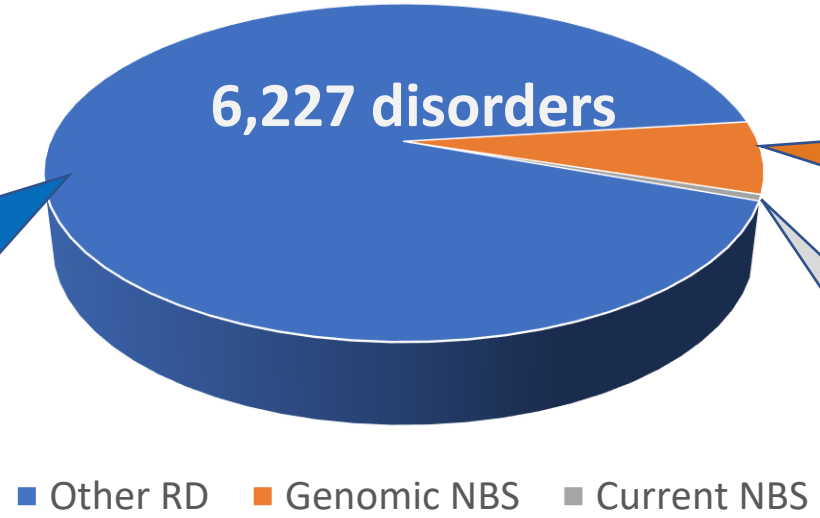
https://www.orphadata.com/

Diagnostic pathways in patients with rare diseases

Diagnosis in clinically symptomatic patients

Clinical care pathway

- Warning signs of RD unrecognized
- Diagnostic odyssey
- Availability of expertise and infrastructure



Pre-symptomatic diagnosis in newborns

Genomic NBS (up to hundreds RD)

- Pilots only
- High hopes from patients
- Risks of adverse effects

Present NBS (up to ≈50 RD)

- Historically proven efficacy
- Biochemical markers/2TT
- Treatable conditions

Stakeholders involved in timely diagnosis and NBS

General public	Payers	Industry
Policy makers	Persons with RD	**Experts**
Researchers	Ethicists and legal experts	**Health data analysts**