

Newborn dried blood spot screening and follow up in Sweden

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Swedish newborn dried blood spot (DBS) screening program

Finding children with severe treatable diseases in which early diagnosis considerably changes prognosis and saves lives

One screening laboratory (CMMS)

115 000 newborns per year (~10 milj inhabitants)

Approx. 100 children found per year

Follow up of approx. 1000 patients using DBS

“PKU biobank” – filter papers stored at +4 since 1975

Ongoing Swedish DBS program (25 diseases)

Two endocrine disorders (CH, CAH)

Paediatric clinic guided by a paediatric endocrinologist in the
NBS laboratory

Genetics for CAH at one site

22 inborn errors of metabolism (IEM)

One of four (soon three) paediatric metabolic centers

Diagnostic workup including genetics at two sites

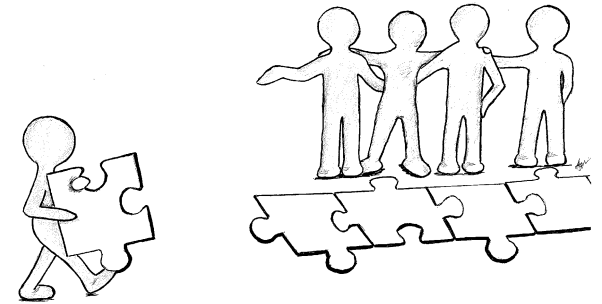


Illustration Jenny Wirgart

One primary immunodeficiency (SCID)

One of three paediatric primary immunodeficiency centers (with HSCT)

Genetics at one site

<https://www.karolinska.se/pku>

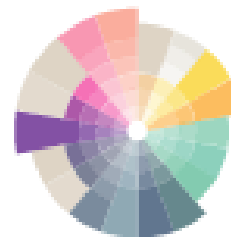
Swedish National Registries



National Swedish Registry for
Inherited Metabolic Diseases

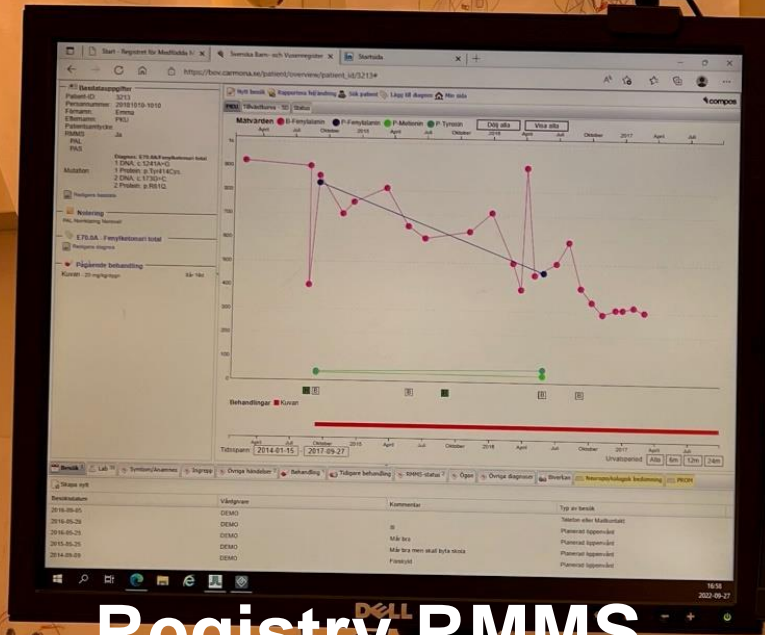
<http://rmms.se>

CAH Registry



**NATIONELLA
KVALITETSREGISTER**

Webcam



Registry RMMS

The Medical Record software interface shows a patient's medical history and current status. The top section is titled 'MOTTAGNINGSENT' and includes fields for patient name, date of birth, and gender. Below this, there is a section for 'Socialt' and 'Aktuella läkemedel'. The 'Socialt' section contains text about the patient's social situation, including mentions of 'Skolan' and 'Sport'. The 'Aktuella läkemedel' section lists 'Kuvan 100 mg 4x1'. The interface also features a sidebar with navigation options and a main content area with a list of medical events and a detailed view of a specific event.

Medical Record



Work station outpatient clinic for IEM Karolinska

CMMS team 2019



Acknowledgements

Photo Rolf Wibom



Svenska Barnläkarföreningen
The Swedish Paediatric Society



ISNS
International Society for Neonatal Screening



European
Reference
Networks

 Socialstyrelsen



Sveriges
Kommuner
och Regioner

And..

CLIR - Collaborative Laboratory Integrated Reports

