ITINERARE Symposium
12th November 2021

The Swiss Rare Disease Registry

PD Dr. phil. nat. **Michaela Fux**, michaela.fux@ispm.unibe.ch

Prof. Dr. med. **Claudia Kuehni**, claudia.kuehni@ispm.unibe.ch

Institute of Social and Preventive Medicine

UNIVERSITÄT BERN

Situation worldwide and in Switzerland

definition

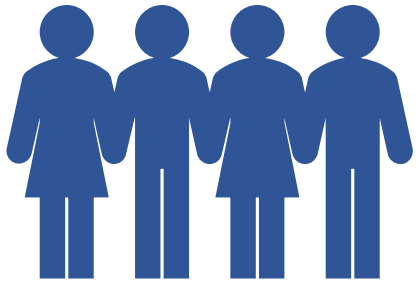
< 1 in 2000

~7000

diseases are classified as **rare**

1 in 12

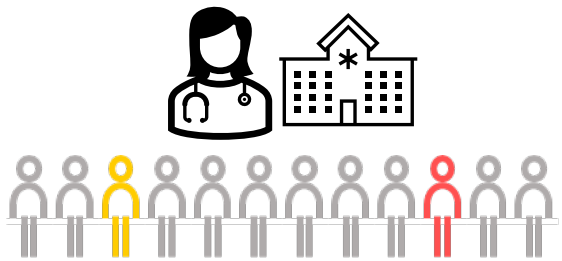
has a **rare disease**

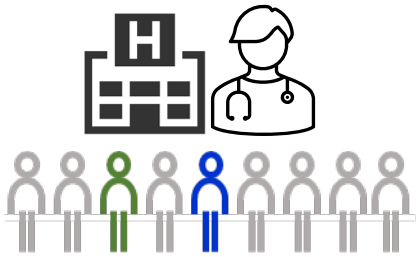


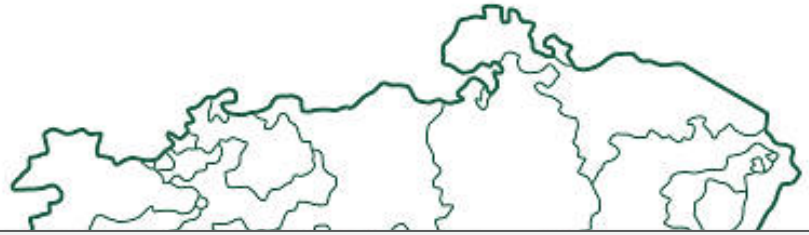
>500 000

persons have a rare disease in **Switzerland**

People with rare diseases are isolated

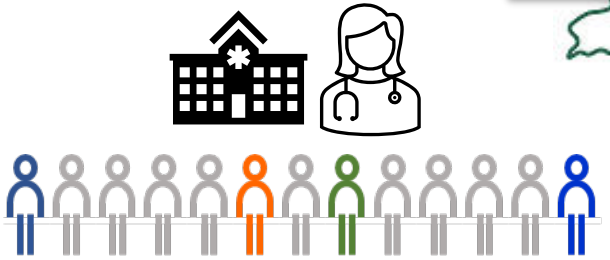






Consequences:

- Affected persons are invisible (ICD)
- Research is impaired
- Medical care is not optimal



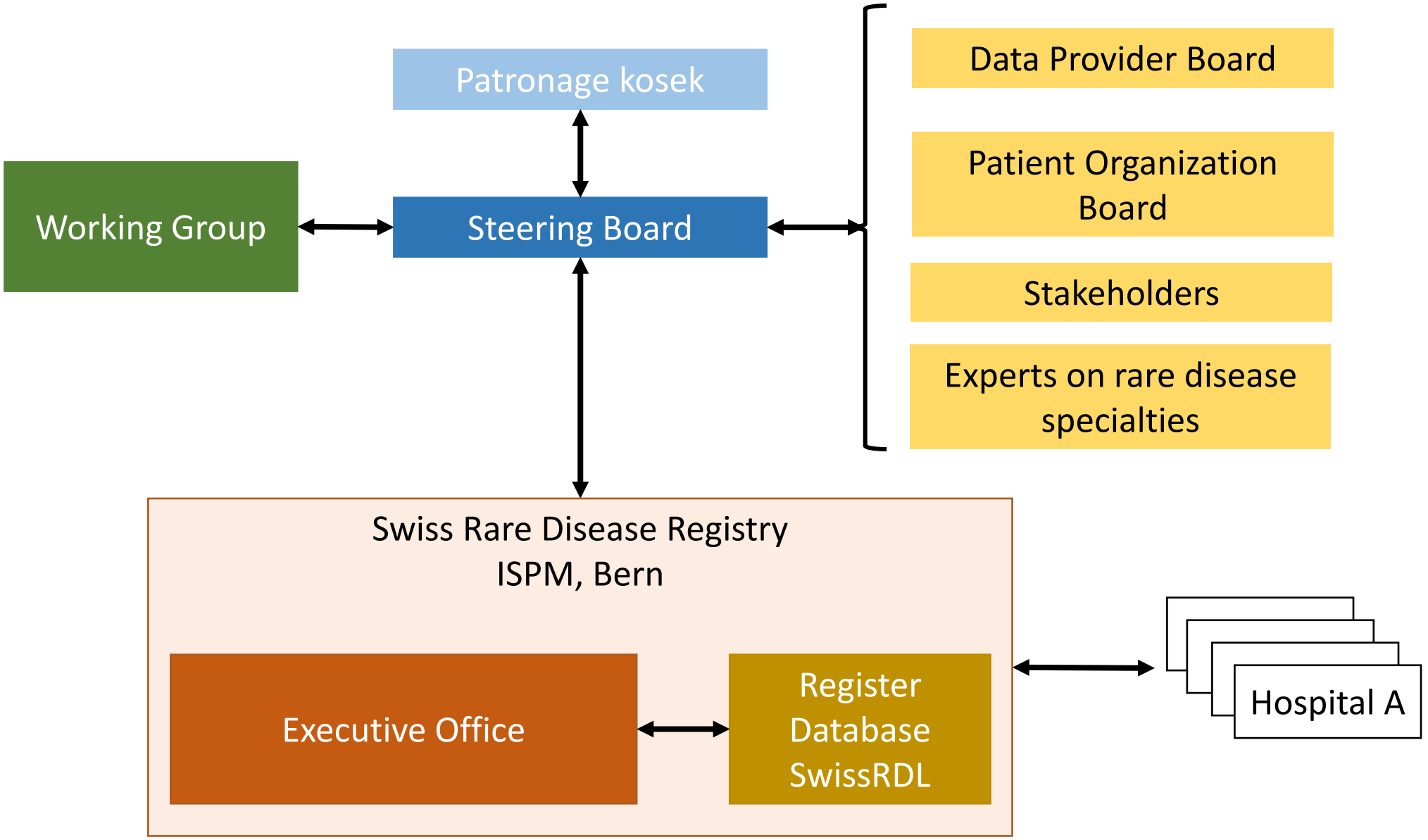
Objectives

- **Setup a research platform** for clinical, epidemiological, basic and translational research on all rare diseases
- **Make people with rare diseases visible in health statistics and research**
- **Describe the situation** of rare diseases in CH
  - **Epidemiology** (incidence, prevalence, survival, mortality)
  - **Health care** (treating institution, diagnostics, management, quality indicators)
- **Facilitate study participation** for patients (national and international)
- **Integrate and harmonize** available data on rare diseases, bundle efforts
- **Build a network for communication**, for patients and health care providers

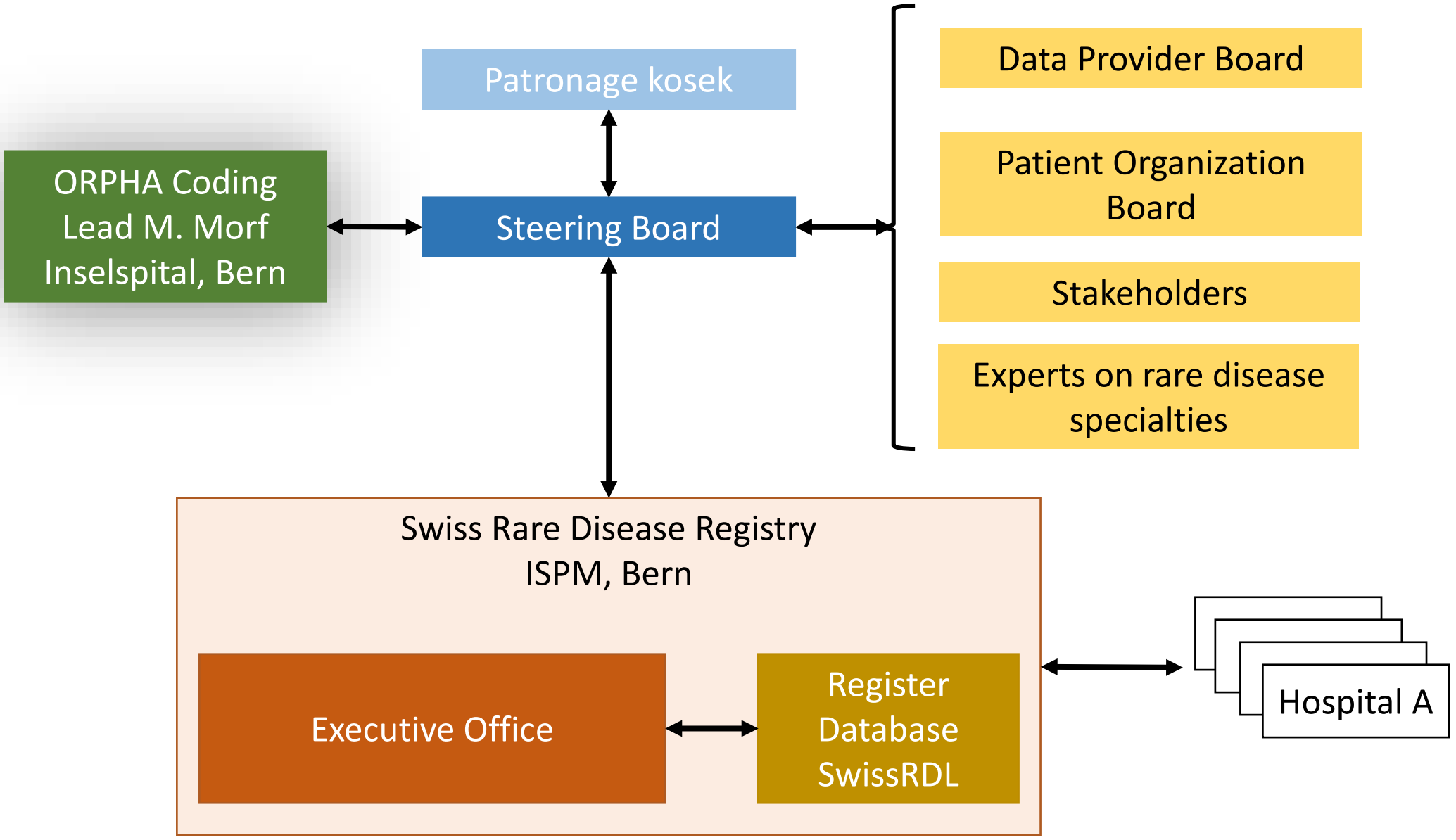
History

2013	• Initiation
2014	• The National Rare Disease Policy
2015-2017	• Concept for the SRDR
2018	• Ethics approval
2020	• FOPH KRG Art. 24 funding approved • (5 years, CHF 250 000 per year) • Further funds needed
2021	• Project Manager PD Dr. Michaela Fux started

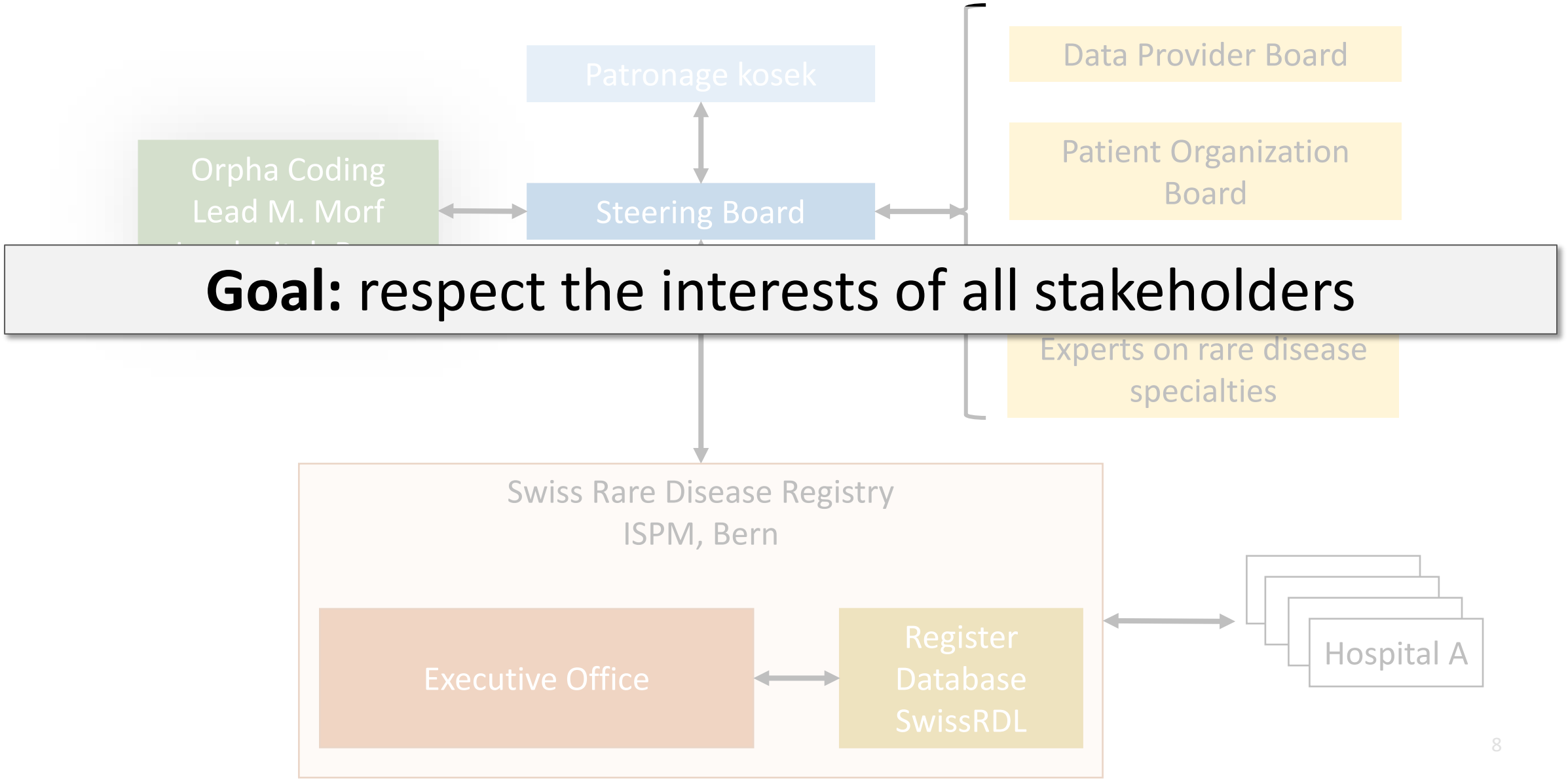
Organization Structure: Concept in development



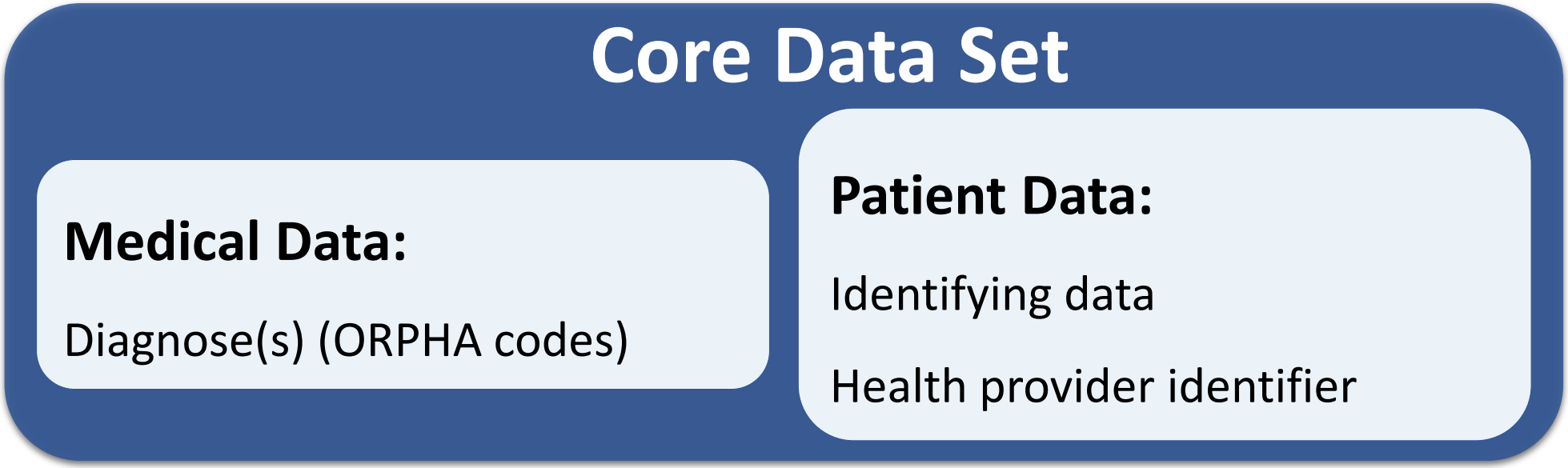
Organization Structure: Concept in development



Organization Structure: Concept in development



Which data do we register?



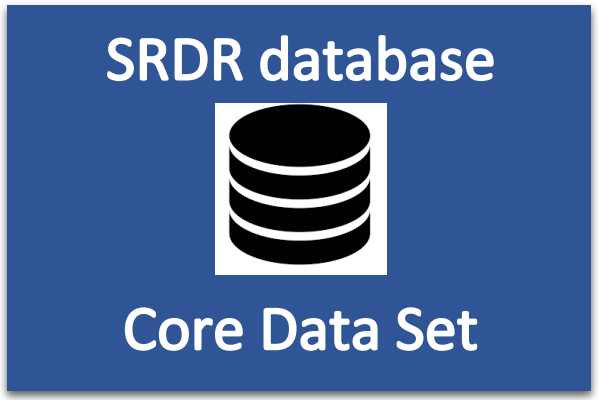
- All data are already in medical records
- **Informed consenting**: required, voluntary and withdrawable

Sources of the Core Data Set

Clinics and practices

Self-registration platform

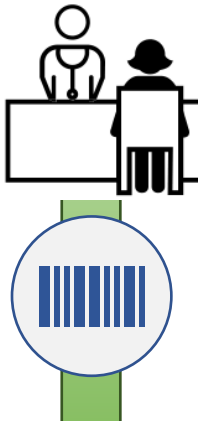
Disease-specific registries



Current focus: Data collection at clinics

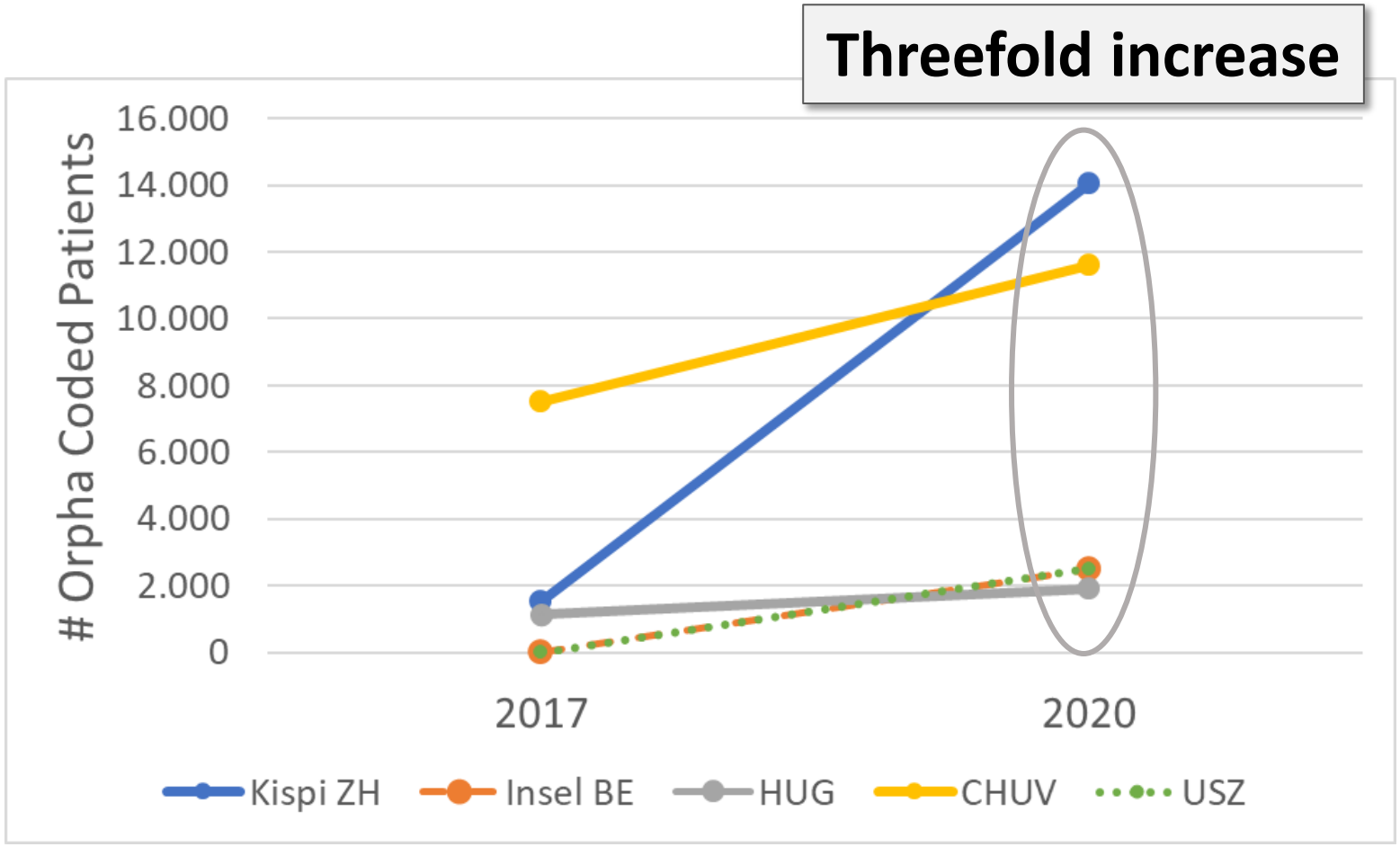
Dx of patient with a rare disease

Orpha Coding



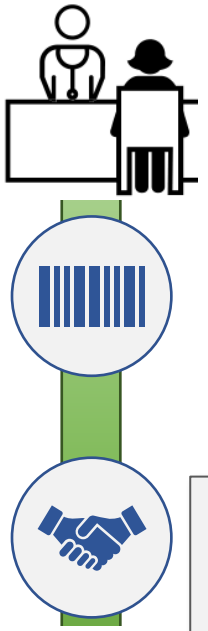
ORPHAcode: a unique and stable identifier for rare diseases

Number of ORPHA coded Patients in Switzerland



Current focus: Data collection at clinics

Dx of patient with a rare disease



Orpha Coding

Information Consenting

- Age-appropriate
- Available in GE, FR and IT
- Clinician and patient sign or only patient signs
- Send to university hospitals, non-university hospitals and patient organizations
- Kispi ZH: 140 signed IC (state July 2021)

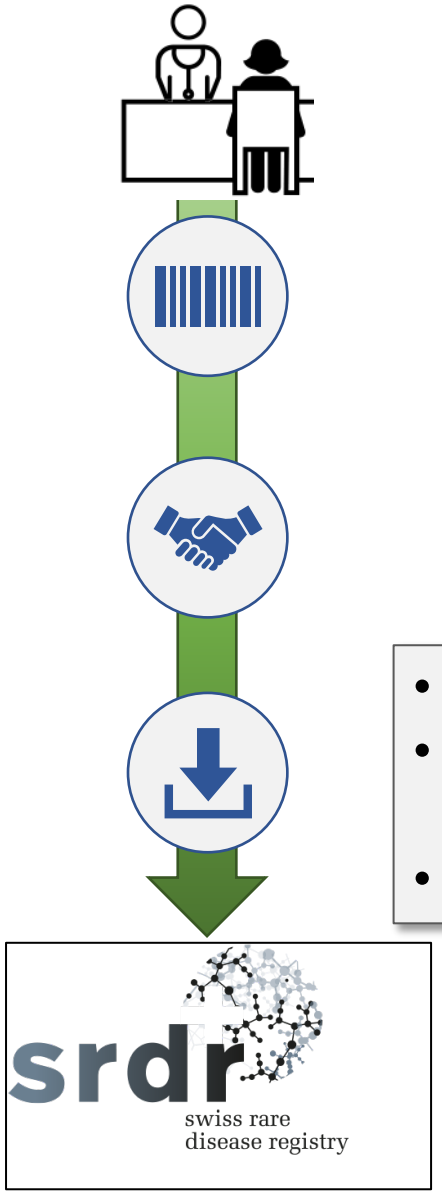
Current focus: Data collection at clinics

Dx of patient with a rare disease

Orpha Coding

Information Consenting

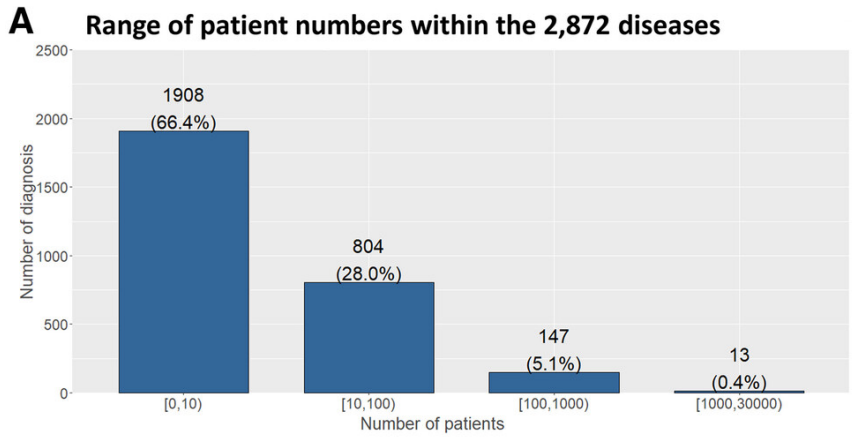
Automatic data transfer

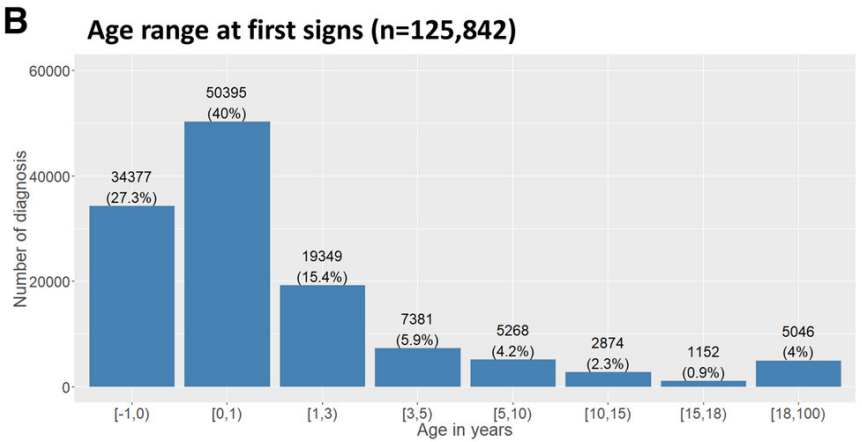


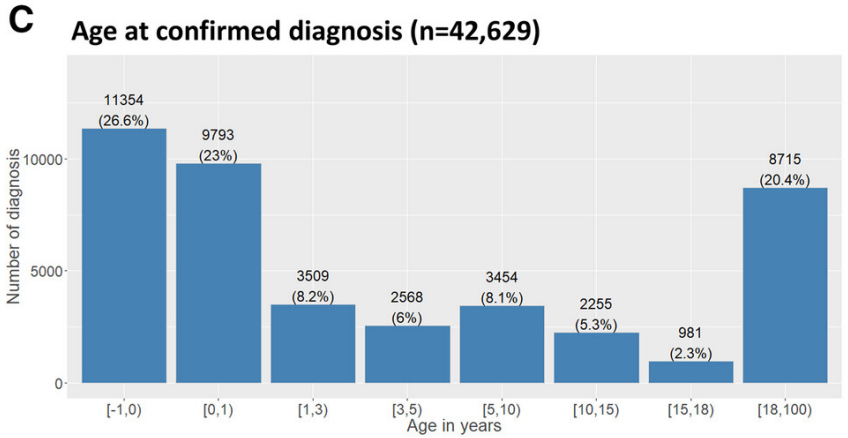
srdr
swiss rare disease registry

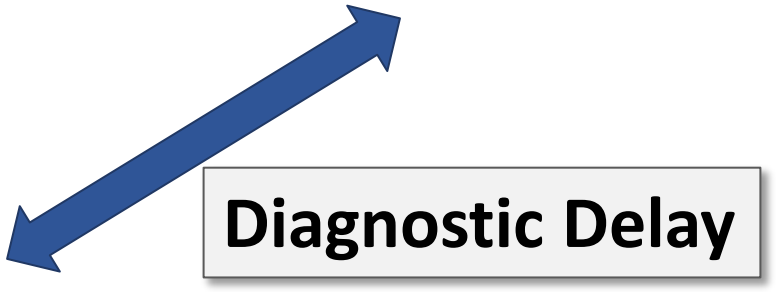
- Process started with BE, GE, ZH Kispi
- Data transfer foreseen for the first quarter of 2022
- More to follow

The Core Data Set: Few, but relevant variables





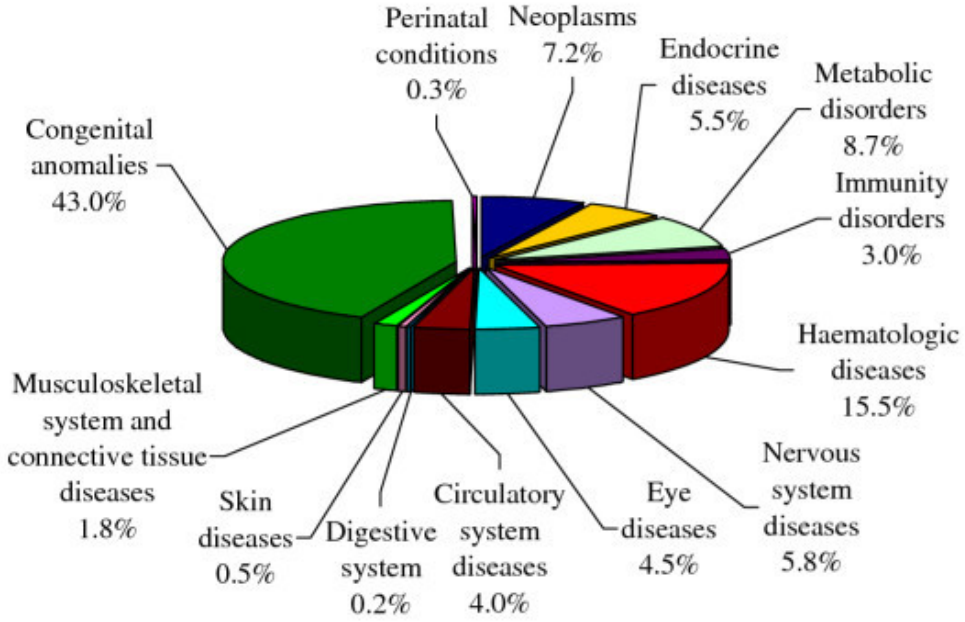




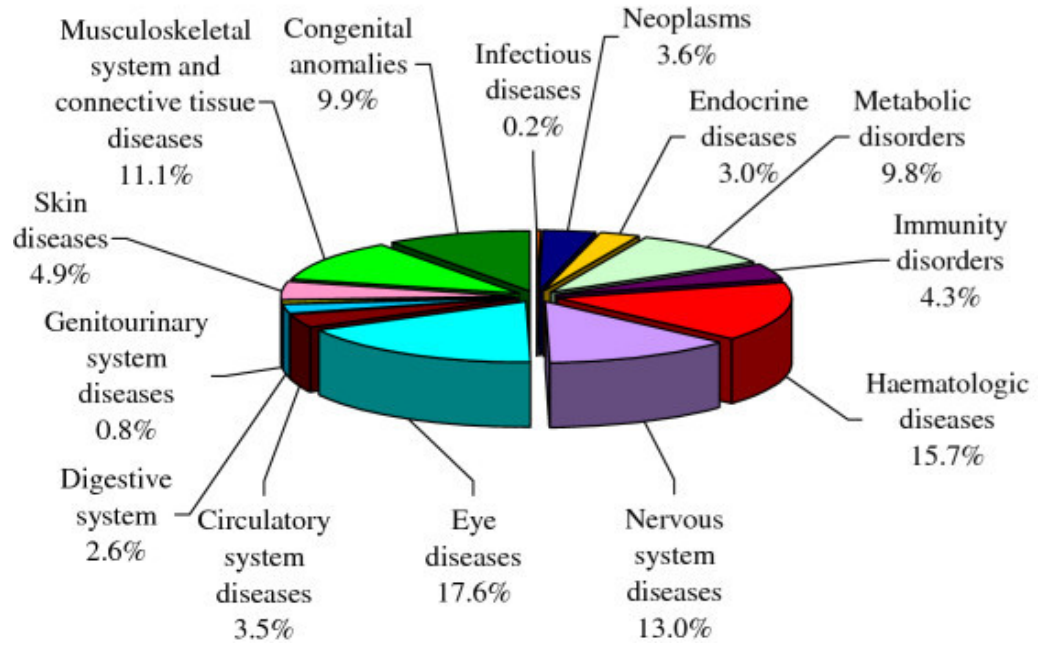
Taruscio D et al, 2014, *Blood Transfus*

The Core Data Set: describing the prevalence…..

0-17 years

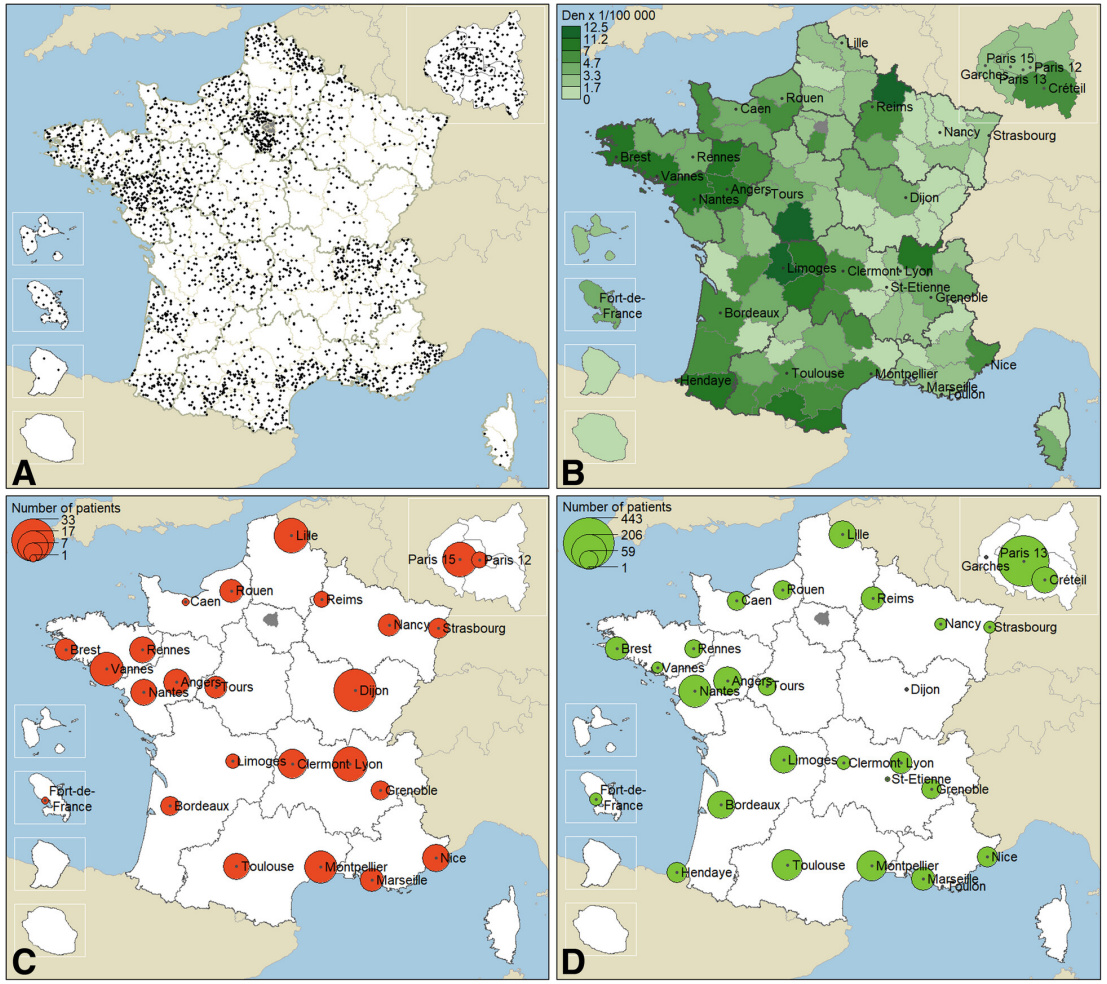


≥18 years



Mazzucato M et al, 2014, *Orphanet J Rare Dis.*

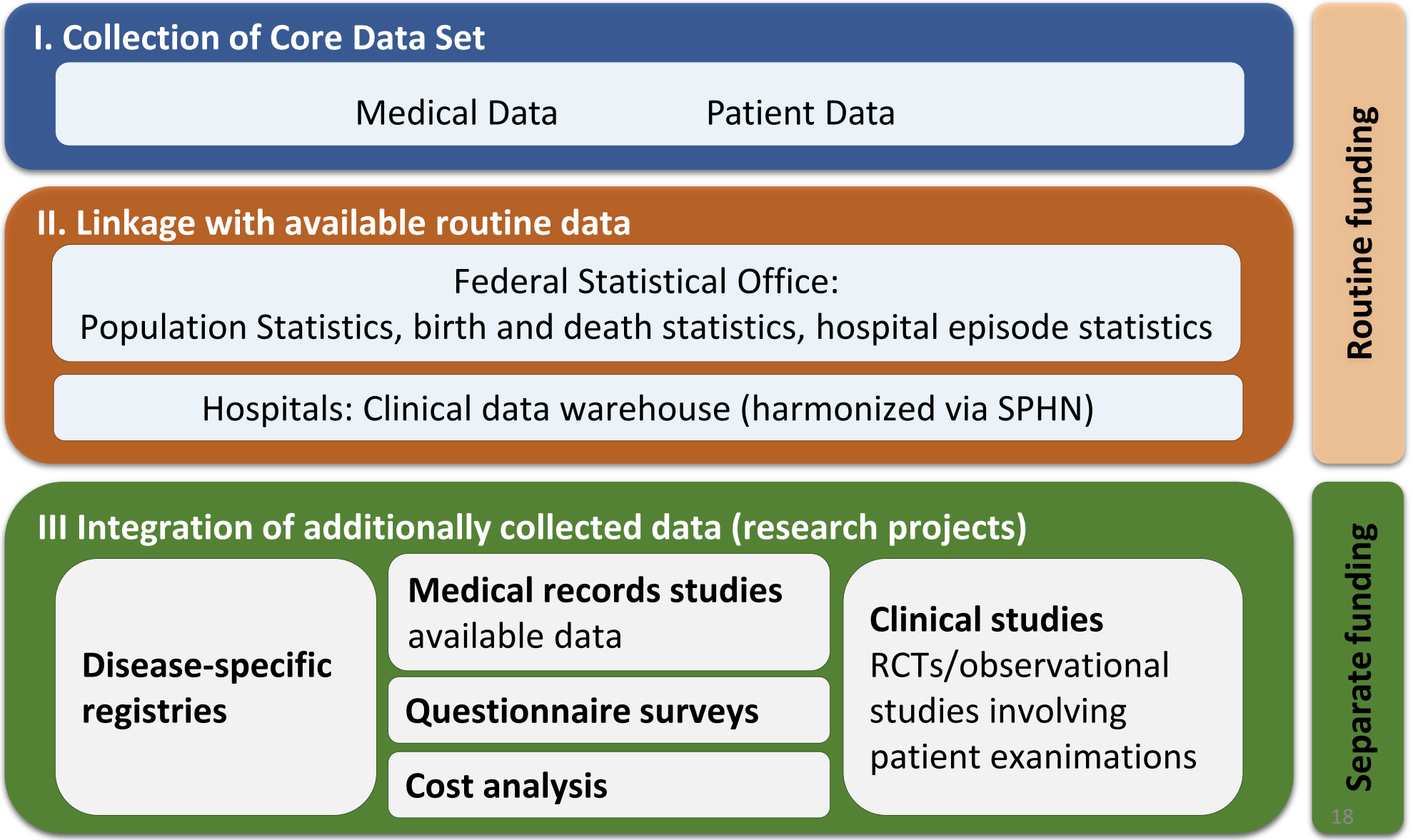
… and the regional distribution



Feasibility study

Antonio, MD et al, 2019,
Orphanet J Rare Dis.

Our design enables diverse studies



Possible studies in SRDR

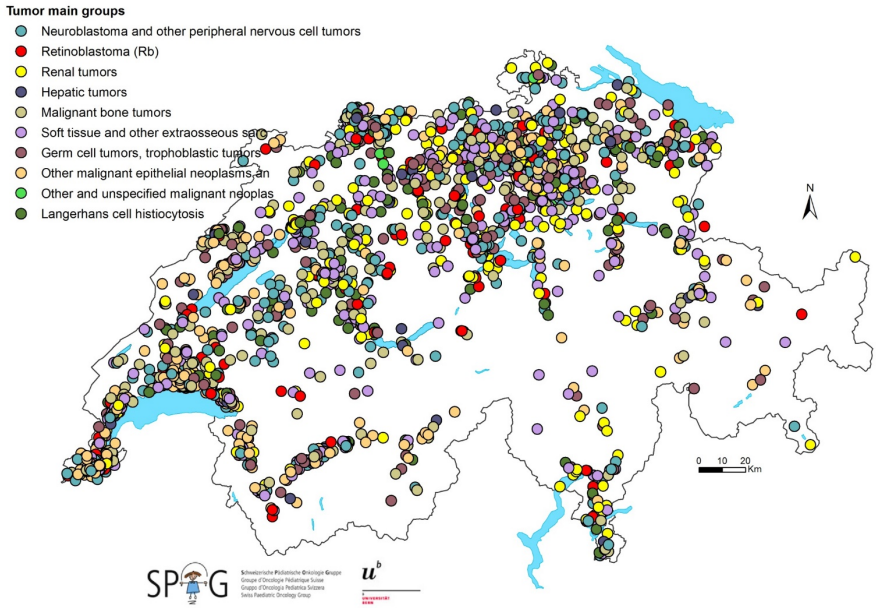
… most examples from Swiss Childhood cancer registry
www.childhoodcancerregistry.ch

Study 1: Prevalence of rare pulmonary diseases + relevant health care providers

Data	**Only core dataset: Diagnosis, age, sex, region**
Work	Extract anonymized dataset from DB Analysis
Ethics	- (included in registry ethics)
Consent	- (included in original consent)
Significance	Insight into prevalence, age structure and regional distribution of diagnosed rare diseases; baseline for other studies

I. Core Data Set

Medical : Diagnosis, date of Dx **Personal**: Age, sex, region, health care



Example from Swiss Childhood Cancer Registry

Study 2: **Survival and causes of death** in patients with rare neuromuscular diseases

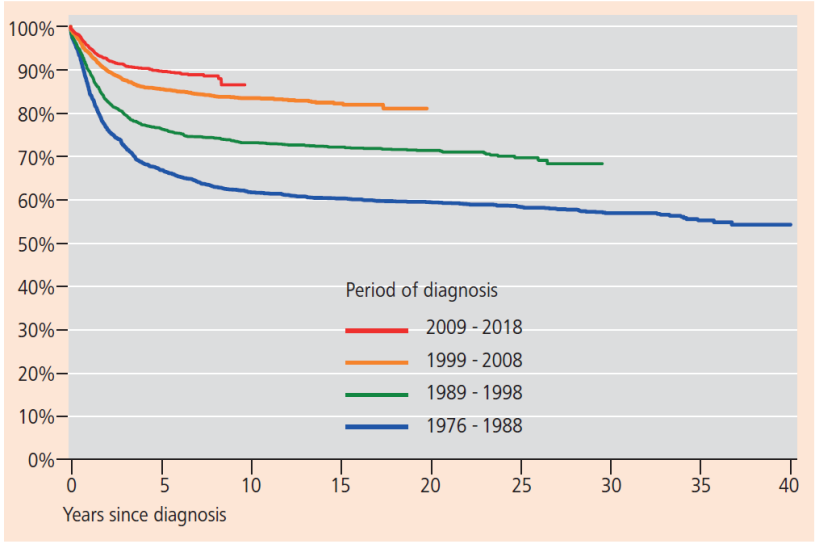
Data	**core dataset** **Cause of death statistics (SFSO)**
Work	Extract dataset from DB Link with dataset from SFSO Analyse pseudonymised dataset
Ethics	**inform** (included)
Consent	- (included)
Significance	Insight into survival of rare disease, trends over time, causes of death, international benchmarking

I. Core Data Set

Medical : Diagnosis, date of Dx **Personal**: Age, sex, region, health care

II. Linked available routine data

Federal Statistical Office: Data and cause of death, birth records, hospitalisations (Dx, duration), geocodable environmental exposures



Study 3: **Air pollution** & risk for interstitial lung disease

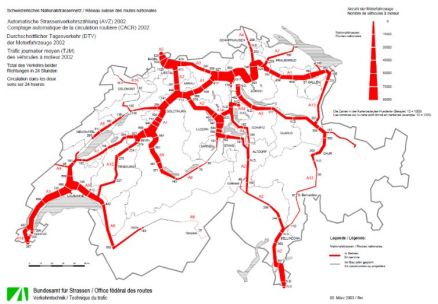
Data	**core dataset** **Geocoded addresses at birth & at dx** **Cohort (healthy peers) from SNC/SFSO** **Exposure data from SFSO, BAFU, etc**
Work	Extract dataset from DB Geocode addresses (cases & peers) Estimate exposure Compare exposure cases : peers
Ethics	inform (included)
Consent	- (included)
Significance	Insight into environmental risk factors for rare disease

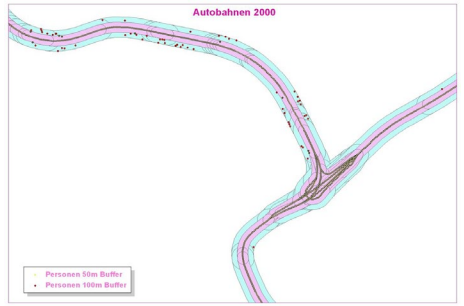
I. Core Data Set

Medical : Diagnosis, date of Dx **Personal**: Age, sex, region, health care

II. Linked available routine data

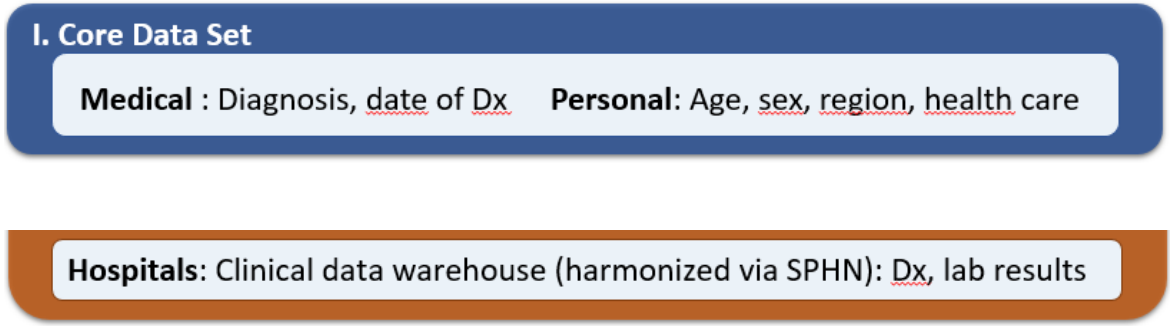
Federal Statistical Office: Data and cause of death, birth records, hospitalisations (Dx, duration), geocodable environmental exposures





Study 4: **Renal function and metabolic syndrome** in patients with …..

Data	**core dataset** **Lab values and blood pressure from university hospital data warehouses (SPHN datasets)**
Work	Extract dataset from DB Link with data warehouses Extract lab values, heights, weights, blood pressure Analyze anonymous dataset
Ethics	- (included)
Consent	- (included)
Significance	Descriptive or analytic studies, long-term course in lab outcomes



Study 5: **Quality of life and PRO outcomes** in patients with .....

Data	**core dataset (Dx & address)** **Additional data collected via questionnaires**
Work	SRDR identifies eligible participants SRDR sends study information & questionnaire Researcher receives anonymous dataset
Ethics	**Small amendment**
Consent	**Collected together with questionnaire**
Significance	Patient-reported outcome measures

I. Core Data Set

Medical : Diagnosis, date of Dx **Personal**: Age, sex, region, health care

Questionnaire surveys


Swiss Survivors

Schweizer Studie zur Gesundheit nach einer Krebserkrankung, Leukämie oder Tumor im Kindes- oder Jugendalter

Fragebogen für Jugendliche

In diesem Fragebogen geht es um die Gesundheit und die Lebensqualität von Jugendlichen Ihres Alters, welche als Kind eine schwere Erkrankung hatten (Krebs, Leukämie oder Tumor). Die Resultate werden den Ärzten helfen, die Behandlung und Nachkontrollen von krebskranken Kindern weiter zu verbessern.

Ihre Antworten werden streng vertraulich behandelt. Die erste Seite und letzte Seite mit Ihrem Namen, Ihrer E-Mail Adresse und Telefonnummer werden von uns vom Fragebogen abgelöst und vertraulich aufbewahrt. Die Eingabe des restlichen Fragebogens in den Computer und die Auswertung erfolgen anonym.

Senden Sie den ausgefüllten Fragebogen bitte im beigelegten und **vorfrankierten Couvert** an uns zurück. Bei Schwierigkeiten oder Fragen dürfen Sie sich gerne an **Dr. med. Rahel Kasteler** oder **Annette Weiss** wenden.

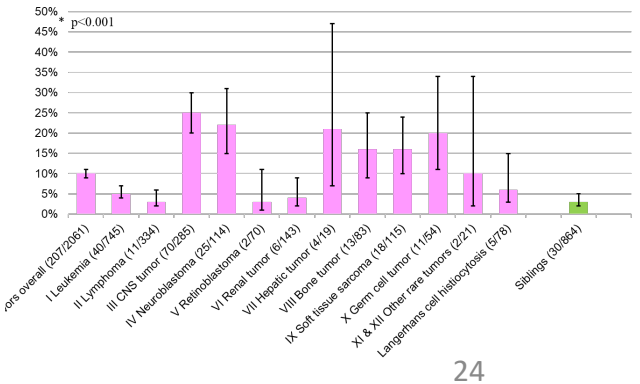
Telefon: 031 631 33 47
E-Mail: rahel.kasteler@ispm.unibe.ch oder annette.weiss@ispm.unibe.ch

bestes dank für ihre hilfe!

Schweizerische Pädiatrische Onkologie Gruppe (SPOG, www.spog.ch)
SPOG Office, Effingerstrasse 40, 3008 Bern

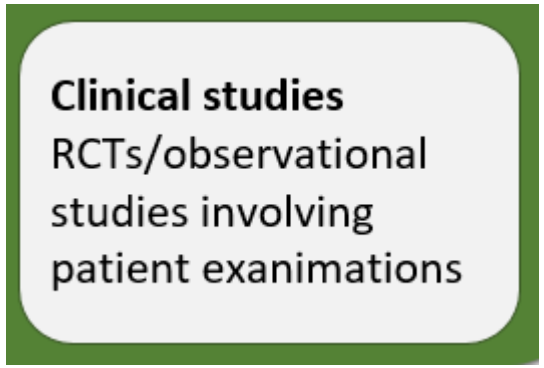
Kinderkrebshilfe Schweiz (www.kinderkrebshilfe.ch)
Florastrasse 14, 4600 Olten

Schweizer Kinderkrebsregister (www.kinderkrebsregister.ch)
Institut für Sozial- und Präventivmedizin, Universität Bern, Finkenhubelweg 11, 3012 Bern



Study 6: **Randomized controlled trial of new drug** X in patients with ….

Data	**core dataset (Dx & address)** **Additional data collected in study**
Work	**SRDR identifies eligible participants** **SRDR sends study info to patients** **Patients** return form to researcher **Researcher** contacts participants (and their physicians) for detailed study information and potential recruitment
Ethics	Separate ethics
Consent	Necessary, oral and written information
Significance	Fast identification & contact of participants for (inter-) national study




Efficacy and safety of azithromycin maintenance therapy in primary ciliary dyskinesia (BESTCILIA): a multicentre, double-blind, randomised, placebo-controlled phase 3 trial

Helene E Kobbernagel, MD • Frederik F Buchvald, PhD • Eric G Haarman, PhD • Carmen Casaulta, MD • Samuel A Collins, PhD • Prof Claire Hogg, MD • et al. Show all authors

Published: May, 2020 • DOI: https://doi.org/10.1016/S2213-2600(20)30058-8 • Check for updates

Study 7: **Set up a patient and family support group** for people living with Noonan syndrome….

I. Core Data Set

Medical : Diagnosis, date of Dx **Personal**: Age, sex, region, health care

Data	**core dataset (Dx & address)**
Work	**SRDR** identifies eligible participants SRDR sends info to eligible people **Interested people can directly contact group organizer /apply 4 membership**
Ethics	Not needed
Consent	Not needed
Significance	Enable people with a rare disease to network with each other

Contact: **Isabelle Cizeau**

In summary

srdr
swiss rare
disease registry

Assuming ..

- sufficient & sustainable funding
- health care providers' willingness and speed of to provide data

The SRDR will be ..

- A national data node and FAIR research platform for all rare diseases
- efficient and affordable

And allows Switzerland to ..

- perform epidemiological studies, health care research & basic research
- Fill gaps in knowledge rapidly
- **Improve health and QOL for people living with a rare disease**