



# Open-Source Registersystem für Seltene Erkrankungen

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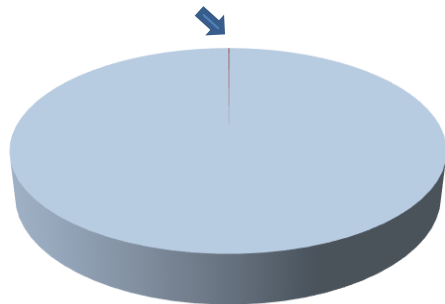
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Datenintegrationszentrum, Dez. 7 - Informations- und Kommunikationstechnologie (DICT), Universitätsklinikum Frankfurt



# Hintergrund: Seltene Erkrankungen

Definition "SE": weniger als **5** Betroffene bei **10.000**.



- > 6,000 Seltene Erkrankungen
- 80% genetischer Ursprung
- 50% betreffen Kinder



- >4 Millionen Betroffene in Deutschland
- > 30 Millionen Betroffene in der EU

[rareiseday.org]

# MIG – Projekte im Bereich SE



**se-atlas** - Versorgungsatlas zur Kartierung von Versorgungseinrichtungen für Menschen mit SE



**ZIPSE** - Zentrales Informationsportal über seltene Erkrankungen



**OSSE** - Open-Source Register-system für Seltene Erkrankungen



**SAVE** - Telemed. Konzept zur Optimierung von Diagn./Therapie v. SE am Bsp. der Cystinose



**NEOCYST** – Network for Early Onset Cystic Kidney Diseases



**ERN LUNG** - Europ. Reference Netw. Lung



**ERDRI** - European RD Registry Infrastructure



**MIRACUM** - Roll Out Seltene Erkrankungen



# Rückblick – Warum OSSE?

- Forderung des Ausbaus der Registerlandschaft, Vernetzung einzelner lokaler Register
  
- **Ziel**  
Einfacher und kostengünstiger Aufbau von Registern, registerübergreifende Nutzung von Forschungsdaten



# OSSE - Motivation

Register



BZ	mg%	99.0
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Seltene Erkrankung X

Register

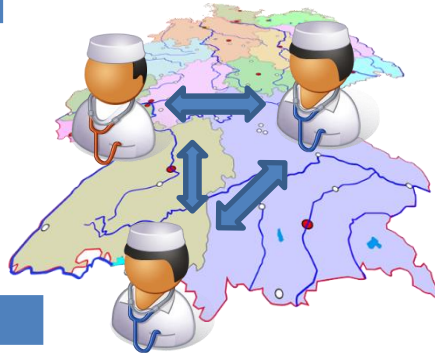


Blutzucker	mg/dl	99
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Register



Glucose im Blut	mmol/l	5,5
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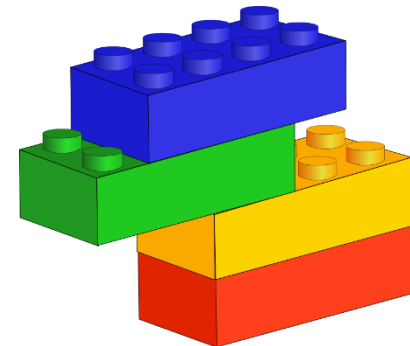


Analyse und Austausch der Daten schwierig!

- Wenige Experten für SE
- Wenige Register
- Wenig Daten
- Unterschiedliche Technologie
  - Papier
  - Tabellenkalkulation
  - Professionelle Datenbanken
  - ...
- Unterschiedliche Datentypen
- Erschwerte überregionale Studien

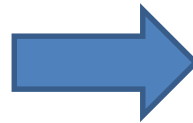
# Einleitung

- **Open Source:** keine Anschaffungskosten für die Software
- Speziell konzipiert als Register für **Seltene Erkrankungen**
- Unterstützung Zusammenarbeit zwischen Registern (**Interoperabilität**)
- **Wahrt den Datenschutz**
- Pseudonymisierung
- Berechtigungskonzept und Zugriffskontrolle
- Datenschutzkonforme Datenrecherche
- Templates: Patienteneinwilligung, Datenschutzkonzept
- Alle Daten bleiben in den Ursprungsregistern, Treuhänder behalten die Datenhoheit

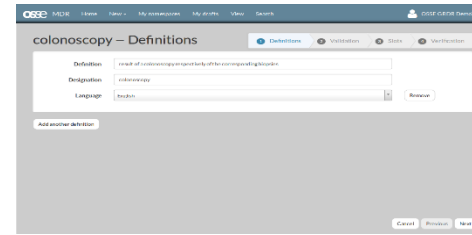


# Erstellen eines OSSE-Registers

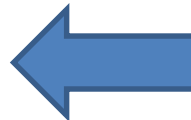
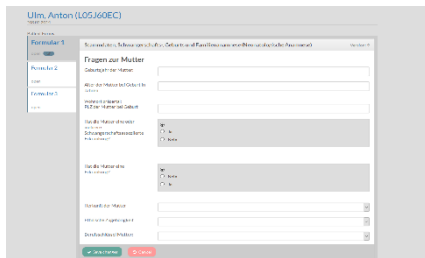
Installation & Konfiguration



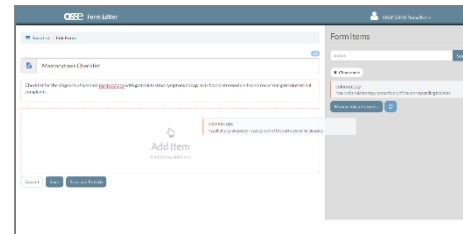
Definition der Datenelemente  
(Metadaten-Repository)



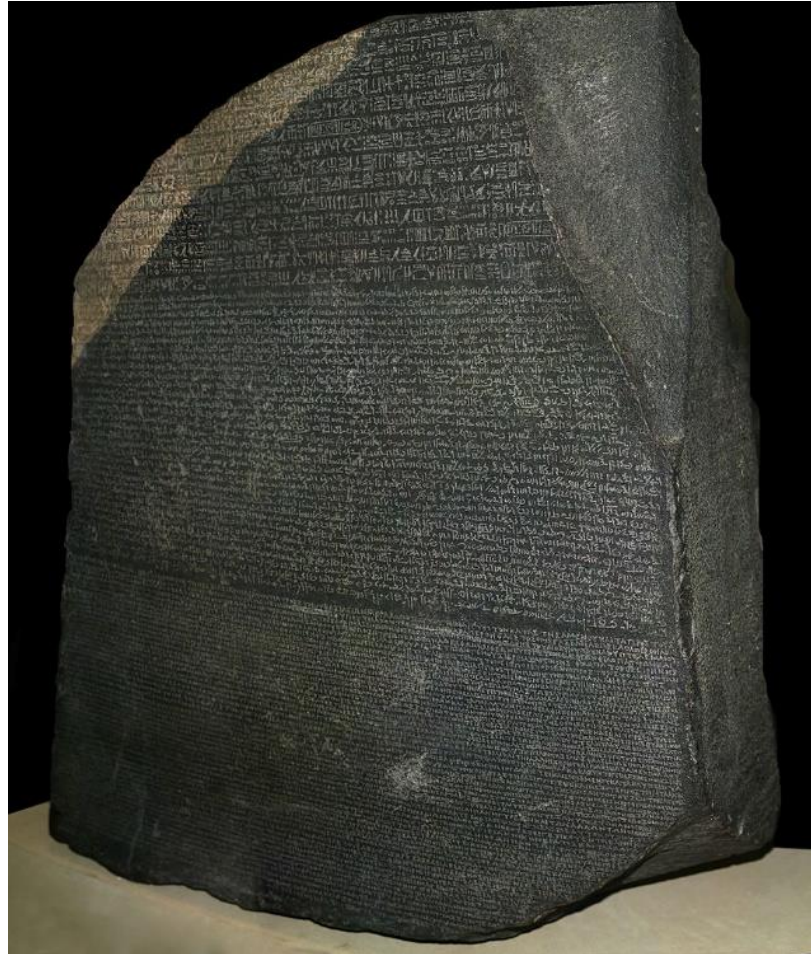
Dateneingabe



Anlegen der Formulare



# Metadaten-Repository



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# Einheitliche Beschreibung von Daten

- Datenelemente werden mit Hilfe von Metadaten definiert (im Metadaten-Repository, *sample.mdr*)



- Beispiel:

- Name **Blutzucker**
- Beschreibung **Unter Blutzucker versteht man im Allgemeinen die Höhe des Glucoseanteils (Glucosespiegel) im Blut.**
- Datentyp **Zahl**
- Einheit **mg/dl**

- Arbeiten mit Standards: ISO/IEC 11179
- Spezifikation entscheidend für Datenqualität



# Erstellen von Datenelementen im Metadaten-Repository

osse MDR Home New ▾ My drafts View Search

Show / Hide Namespaces

Filter

My namespaces

osse-131 : ECLIP  
ECLIP

Other namespaces

mdr3 : ISO 3166-1 Alpha 2  
ISO 3166-1 Alpha 2 ist ein Standard für die zweistellige Kodierung von Staaten, herausgegeben von der Internationalen Organisation für Normung (ISO).

adt : ADT  
Der einheitliche onkologische Basisdatensatz der ADT und der GEKID

osse-97 : AMLREG  
AMLREG

## Browse

▼ adipocytes/preadipocytes tiss  
adipocytes/preadipocytes tissue

▼ Blood pressure left arm (diast  
Blood pressure left arm (diastolic

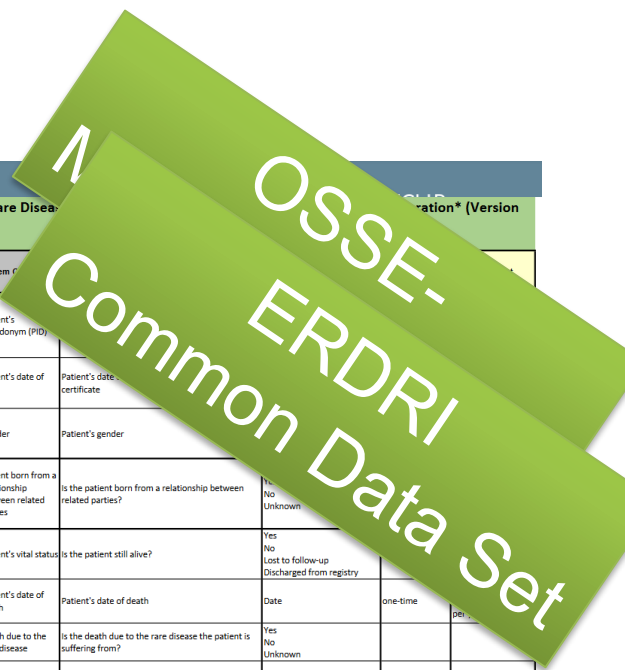
▼ C3 complement  
C3 complement

▲ Large mouth  
Large mouth

Validationtype List of permitted values

Permitted value	Language	Designat
1	EN	Yes
2	EN	No
3	EN	Unknown

Item Group	Item No.	Item Name	Description	Value Type	Cardinality	Units	Other
2. Personal Information 1.. Pseudonym	1.1	Patient's pseudonym (PID)					
	2.4	Patient's date of birth	Patient's date of birth certificate				
	2.5	Gender	Patient's gender				
3. Family Information	3.1	Patient born from a relationship between related parties	Is the patient born from a relationship between related parties?	Yes No Unknown			
	4.1	Patient's vital status	Is the patient still alive?	Yes No Lost to follow-up Discharged from registry			
4. Vital status	4.2	Patient's date of death	Patient's date of death	Date	one-time		
	4.3	Death due to the rare disease	Is the death due to the rare disease the patient is suffering from?	Yes No Unknown			
	5.1	Patient's date of inclusion in the registry	Date at which the patient was included in the registry	Date			
6. Disease history	6.1	Age at onset	Age at which symptoms first appeared	Antenatal At birth XX year(s) and XX month(s) Undetermined			
	6.2	Age at diagnosis	Age at which the diagnosis was made	Antenatal At birth XX year(s) and XX month(s) Undetermined			
7. Diagnosis	7.2	Diagnosis of the rare disease	Diagnosis retained by the RD center	Alpha code			
8. Research	8.1	Agreement to be contacted for a protocol	Does the patient give permission to be contacted for a research protocol?	Yes No			
	8.2	Patient non-opposition to the reuse of data	Does the patient give permission for his/her data to be reused for other research purposes?	Yes No			
	8.3	Patient having previously given a biological sample for research	Has the patient already given a biological sample for research?	Yes No			
	8.4	Patient having previously given a biological sample for molecular diagnosis	Has the patient already given a biological sample for molecular diagnosis?	Yes No			





# Formular-Editor: Beispiel Ohne-Diagnose Register

OSSE Form Editor
Ohne Diagnose Register

Form List / Edit Form

Vital Status v7 published

Vital Status

Patient's vital status   Repeatable  Mandatory

Patient's date of death   Repeatable  Mandatory

Death due to the rare disease   Repeatable  Mandatory

Gender   Repeatable  Mandatory

Patient's gender   Repeatable  Mandatory

Death due to the rare disease   Repeatable  Mandatory

Further Symptoms

Diagnosis Code (Symptom)

Type of code (Symptoms)

Symptom ontology description   Repeatable  Mandatory

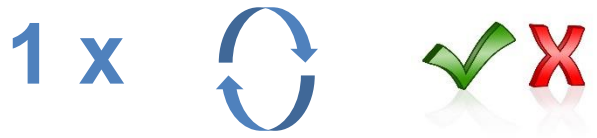
Symptom priority

Year of first manifestation

Form Items

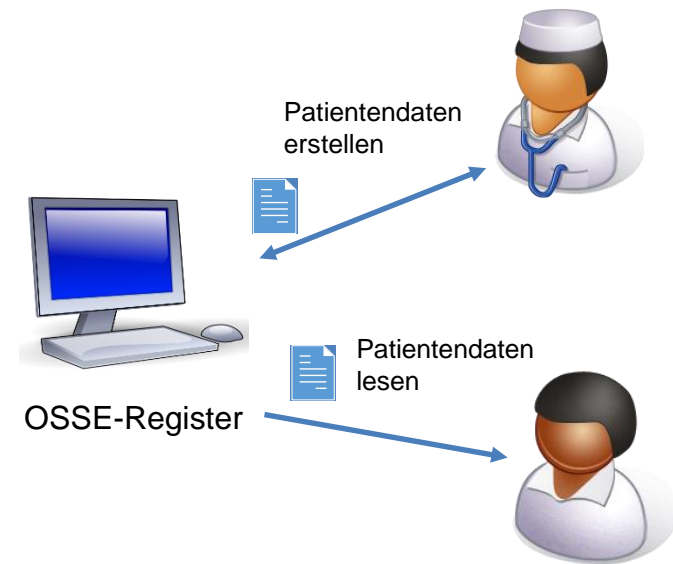
Design Elements

- Gender  
Patient's gender
- Patient born from a relationship between related parties  
Is the patient born from a relationship between related parties?
- Death due to the rare disease  
Is the death due to the rare disease the patient is suffering from?
- Agreement to be contacted for a protocol  
Does the patient give permission to be contacted for a research protocol?
- Patient non-opposition to the reuse of data  
Has the patient been informed concerning and not opposed to a future re-use of his/her de-identified data for other research purposes?
- Patient having previously given a biological sample for research  
Has the patient already given a biological sample for research?
- Patient having previously given a biological sample for molecular diagnosis  
Has the patient already given a biological sample for molecular diagnosis?
- Stage of life  
Stage of life
- Age in years  
Age in years
- Age in months  
Age in months
- HPO-Code  
HPO-Code
- Description of the chosen HPO-code  
Description of the chosen HPO-code
- Date of first manifestation  
Date of first manifestation





# Rollen – und Benutzerkonzept


- Unterschiedliche Lese- und Schreibzugriffe
  - Hinzufügen neuer Patienten und Eingabe medizinischer Daten
  - Lesen und Schreiben medizinischer Daten
  - Datenexport
  - Standorte verwalten
  - .....





# Erfassung von Patientendaten

OSSE  List of patients UK  Jannik Schaaf

 ( ABCD123 )

Episodes (8)

31/10/2013 First Visit	27/11/2013 Visit 2	27/12/2013 Visit 3	10/02/2014 Visit 4	10/04/2014 Visit 5	08/07/2014 Visit 6	01/09/2014 Visit 7	11/12/2014 Visit 8
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Episode: 31/10/2013

<p>Anthropometric data</p> <p>open</p> <p>Skinfolds</p> <p>open</p> <p>Distribution of lipodystrophy</p> <p>open</p> <p>Dexa Analysis</p> <p>open</p> <p>Organ Systems (Skin &amp; Appendages)</p> <p>open</p> <p>Organ Systems (Skull &amp; Face)</p>	<p>Organ Systems (Hematology) <span style="float: right;">Version: 3</span></p> <p>Organ Systems (Hematology)</p> <p>Hematology <input type="text" value="Abnormal"/></p> <p>Anaemia <input type="text" value="Yes"/></p> <p>Coagulation abnormalities <input type="text" value="Yes"/></p> <p>Increased erythrocyte sedimentation rate <input type="text" value="No"/></p> <p>Hypergammaglobulinemia <input type="text" value="Unknown"/></p>
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# Lokale Suche / Export / Anbindung

- Suche in eigenem Datenbestand
- Export-Funktion (z.B. Excel)
  - Auswertungszeitraum
  - Filtermöglichkeiten
  - Auswahl der Datenelemente
- Schnittstellen
  - REST-Schnittstelle
  - FAIR-Data-Point

The screenshot shows the 'Data export' interface of the OSSE Share system. At the top, there are navigation tabs: 'OSSE.Share', 'Decentral Search', 'Data export' (selected), and 'Charts'. An 'exporter' dropdown is visible in the top right corner.

The main content area is titled 'Data export' and includes a 'Filter by date' section with three rows of date range filters: 'Changed between', 'Created between', and 'Episode date between'. Each filter has two input fields and a calendar icon, with an 'and' operator between them.

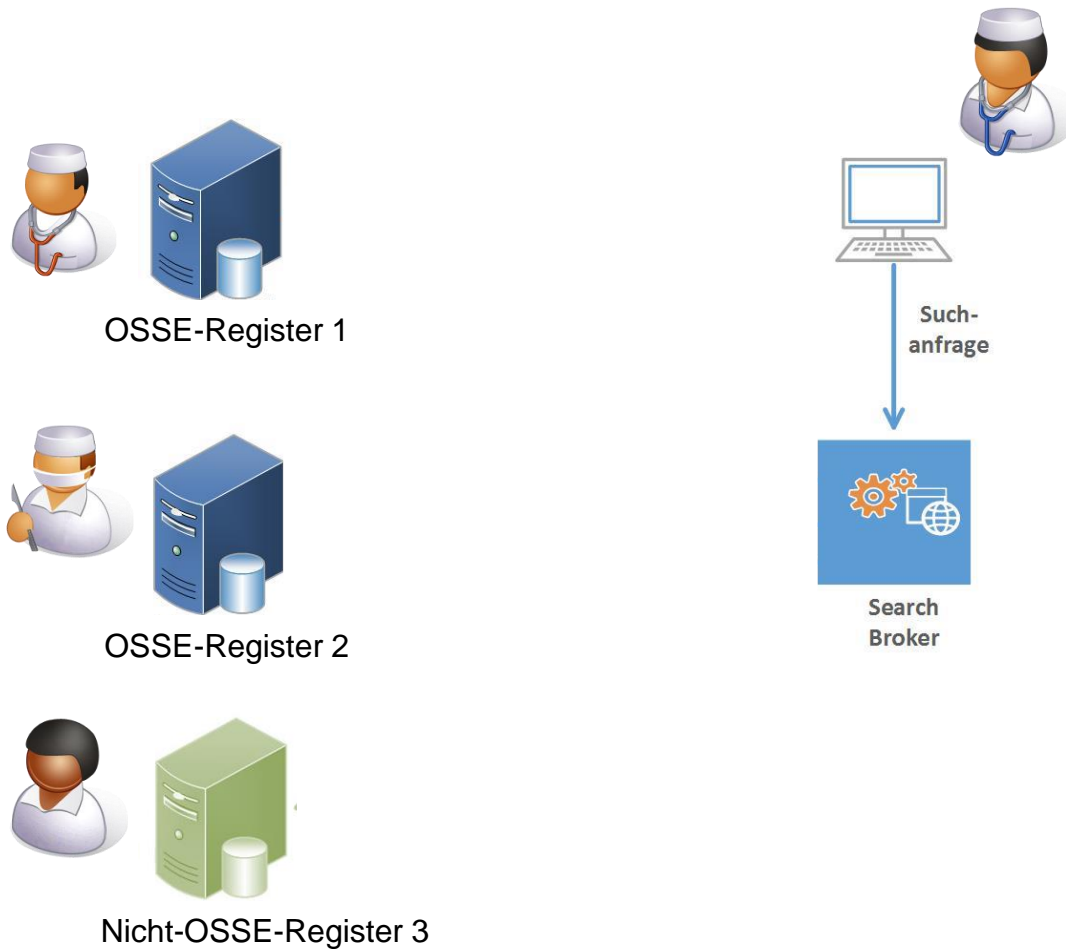
Below the filters is the 'Search criteria' section, which includes an 'Edit search criteria' button.

The 'Items to export' section features a refresh icon and a list of exportable items, each with a checkbox and a version indicator (v.1):

- Check all/Uncheck all
- PLZ der Mutter bei Geburt  
PLZ der Mutter bei Geburt [0-99999]
- Gravida  
Wie oft Schwanger gewesen?
- Para  
Wie oft ein Kind geboren?
- Freitext  
Freitext
- Davon spontane Frühaborte (<12 SSW)  
Davon spontane Frühaborte (<12 SSW)
- Davon spontane Spätaborte (>12 SSW)  
Davon spontane Spätaborte (>12 SSW)
- Davon induzierte Aborte  
Davon induzierte Aborte
- Verstorben nach Lebensmonaten  
Verstorben nach Lebensmonaten
- Risikonummern laut gelbem Heft  
Risikonummern laut gelbem Heft

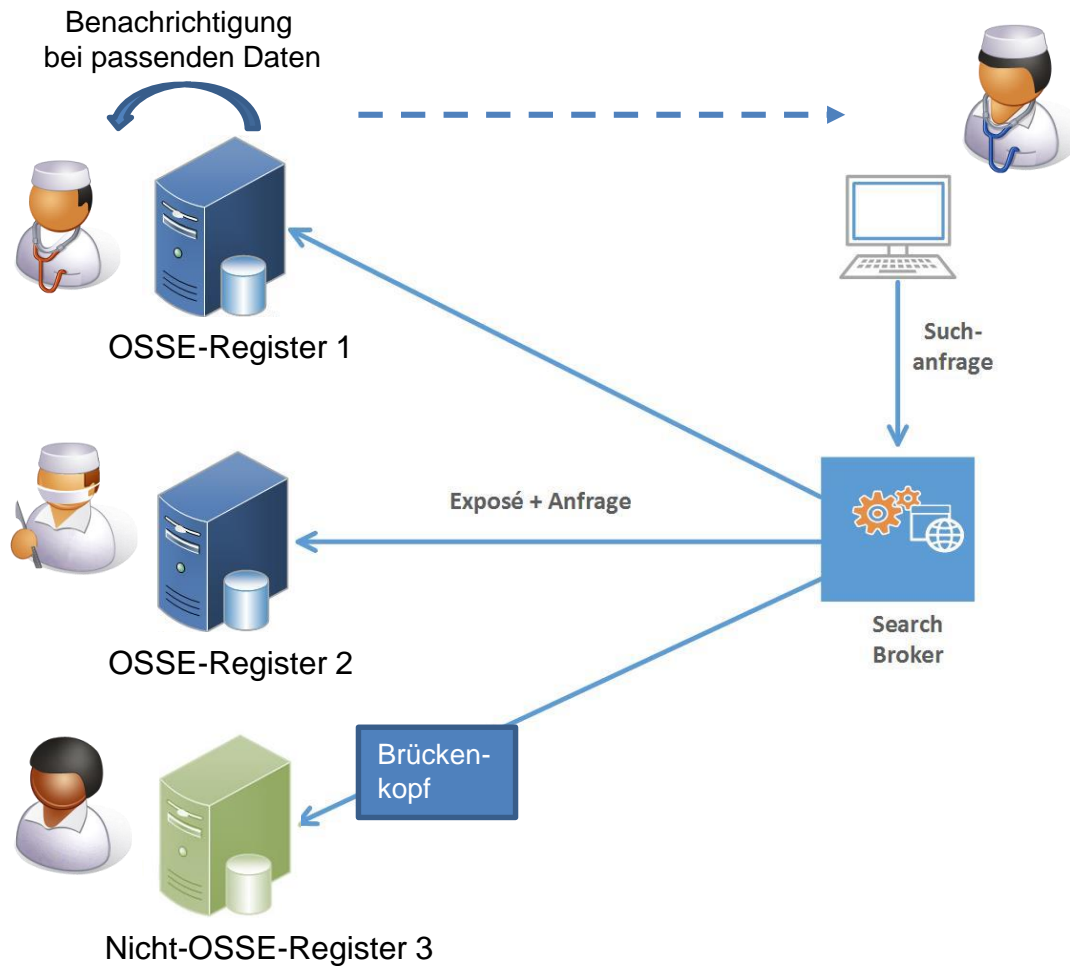


# optionale Zusammenarbeit über „verteilte Suche“





# optionale Zusammenarbeit über „verteilte Suche“





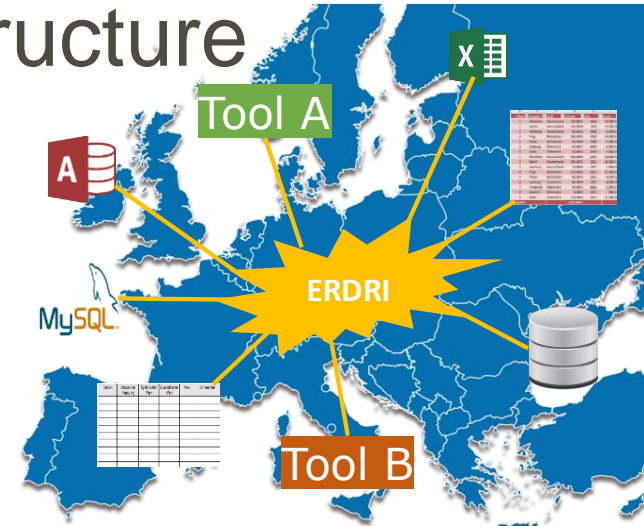
# Bisherige Installationen

- Ohne Diagnose Register, *FRZSE Frankfurt*
- Mukoviszidose-Register (Brückenkopf), *Uniklinik Freiburg*
- NEOCYST-Register für zystische Nierenerkrankungen  
*Konsortium: UK Münster, UK Köln, UK Duisburg-Essen, MH Hannover, UK Heidelberg, Zentrum für Humangenetik Bioscientia Ingelheim, UK Freiburg*
- Marfan-Register, *Uniklinikum Hamburg-Eppendorf*
- Register für seltene angeborene Fehlbildungen, *Kinderklinik Frankfurt*
- Stammzelltransplantationsregister, *Kinderklinik Frankfurt*
- Onko-Fertilitäts-Register, *Kinderklinik Frankfurt*
- Register für kraniofaziale Fehlbildungen (im Aufbau), *Uniklinik Münster*
- TEVAR Register, *Uniklinikum Hamburg-Eppendorf*
- Cystinose-Register, *SAVE-Konsortium*
- Von-Hippel-Lindau-Register, *Selbsthilfe VHL*
- Europäisches Lipodystrophie-Register, *Uniklinikum Ulm*
- + Europäische Referenznetzwerke  European Reference Networks
- + C4-Projekt

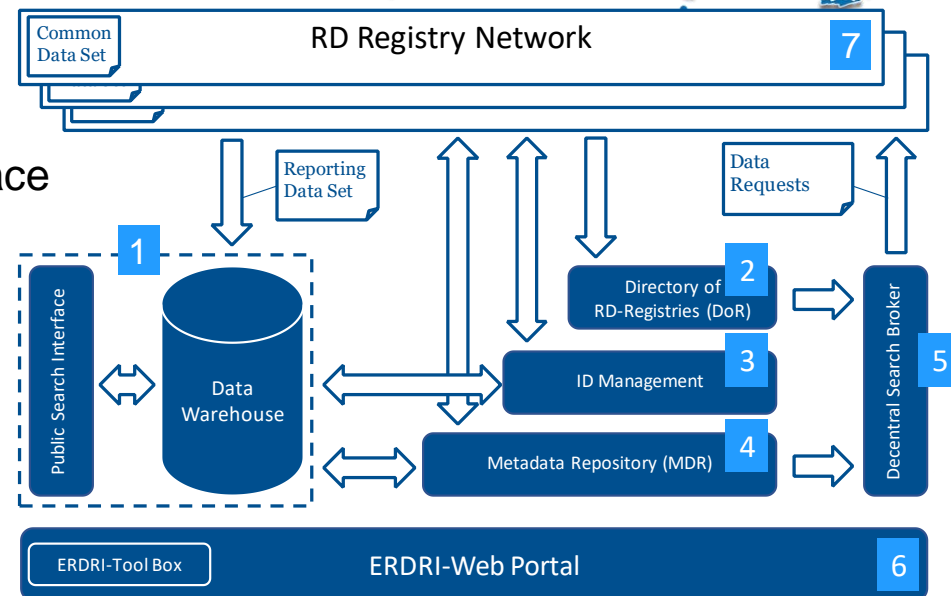


# European RD Registry Infrastructure

- In Kooperation mit dem Joint Research Centre (JRC) der EU



- 1 Data Warehouse with Public Search Interface
- 2 Directory of RD-Registries (DoR)
- 3 ID Management
- 4 Metadata Repository (MDR)
- 5 Decentral Search Broker
- 6 ERDRI Web Portal with Tool Box
- 7 RD Registry Network





Acknowledgements

# Vielen Dank



www.mig-frankfurt.de

www.osse-register.de

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