Láthatóság

Módszertani korlátok

„Monitoring-prevenció”

Coding of rare diseases in the National Health Insurance Fund discharge recording

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Workshop on coding of rare diseases in health information systems with Orphacodes
EC Joint Research Centre, Ispra, 1-2 October 2014

OEP teljesítmény-elszámolási rekordok

- Teljes szakellátást lefedi
- Finanszírozási adatbázis
- Jogi keretek kidolgozottak
- Informatikai és adminisztratív eljárások
- Standard rekordok, ellenőrzés, korrekciós eljárás

- Rekordba beilleszthető fizikailag több kód
- Potenciális adatbázis (szükségletelemzés, surveillance, minőségbiztosítás)

ORPHA kódok az OEP teljesítmény-elszámolási rekordjaiban

RBK

OEP

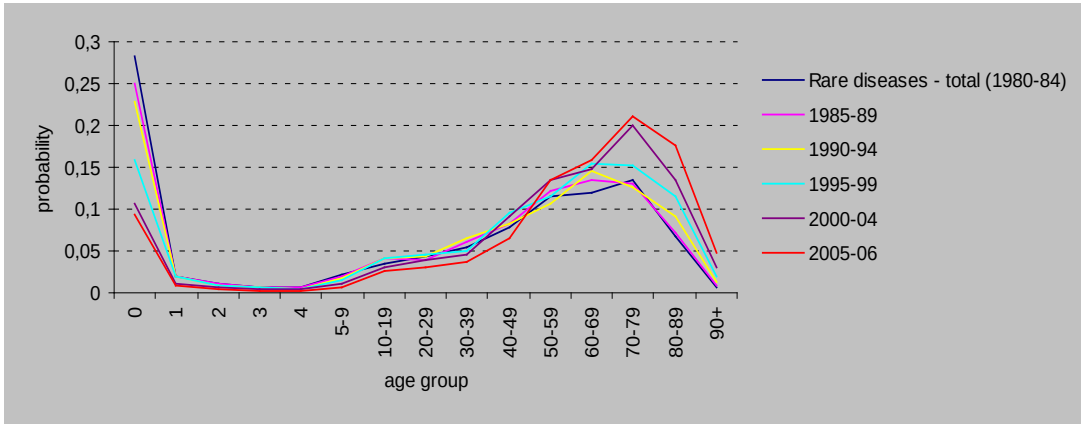
Államtitkárság

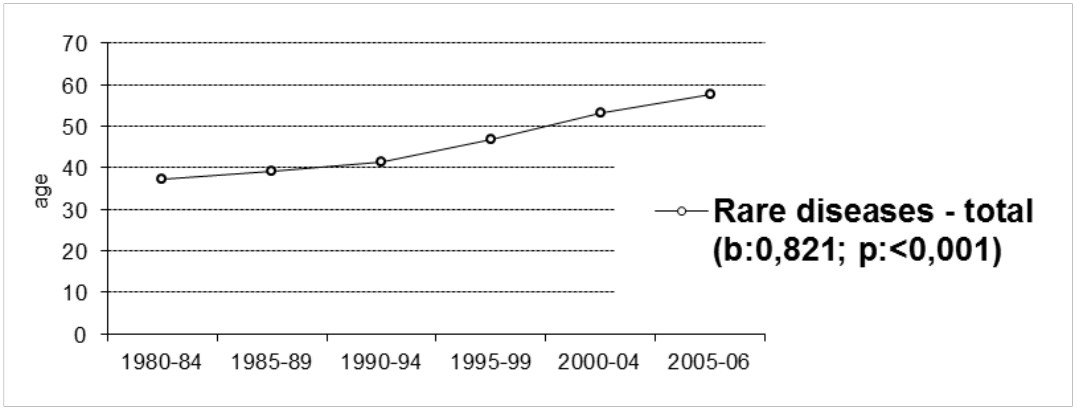
- Kötelező ORPHA kódolás a szakellátásban
- *Előre definiált BNO10 kódok csak ORPHA kód deklarációval fogadhatók be*

ORPHA kódok az OEP teljesítmény-elszámolási rekordjaiban

1. RD-specifikus adatok gyakorlati értékének demonstrálása:
   – Okspecifikus halálozás
2. RD specifikus OEP indikátorok gyakorlati értékének demonstrálása:
   – Népegészségügyi indikátorok
   – Ellátás minőség indikátorai
3. RD-specifikus kódok kapcsolásának technikai lehetősége

1. Rare diseases mortality, Hungary (1980-2006; N=44775)





For the studied RDs:

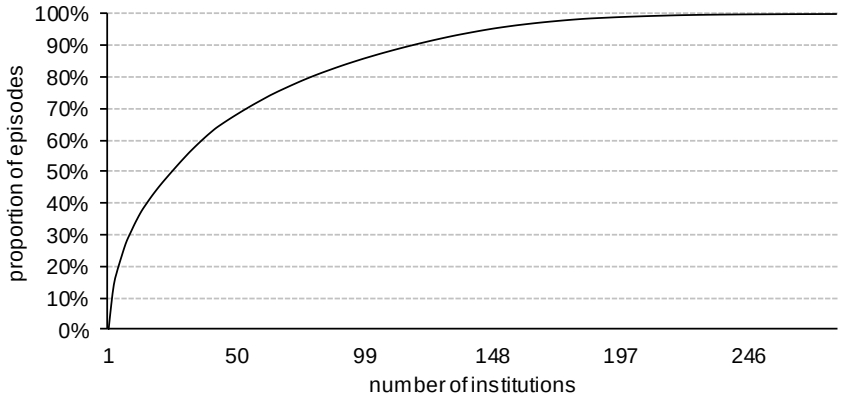
- Remarkable improvement
- Changed social representation of RD

1. Rare diseases mortality, Hungary (1980-1984; 2005-2006)

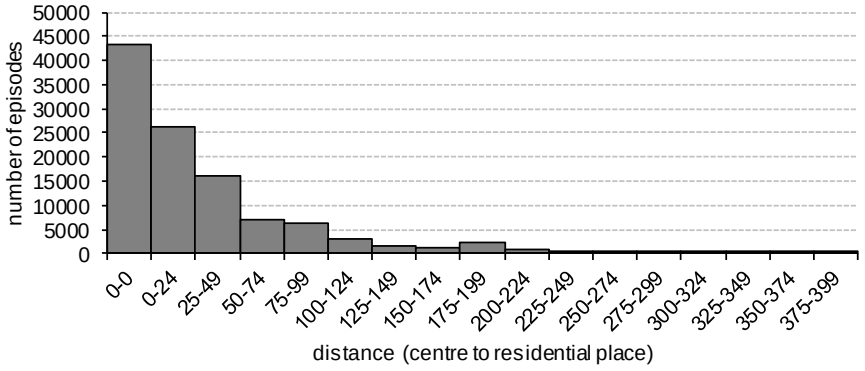
Indicators	1980-1984	2005-2006
Registered standardized death rate (SDR/1,000,000)	**208**	**141**
Proportion of death during the first year of life [95%CI]	**0,28 [0,27 - 0,29]**	**0,09 [0,08 - 0,10]**
Mean age at death attributed to rare diseases (p for change < 0.001)	**37,3**	**57,7**
Potential years of life lost attributed to rare diseases (PYLL)	**66119**	**21900**

2. RD patient pathway analysis, HDR study (Hungary, 2004-2009)

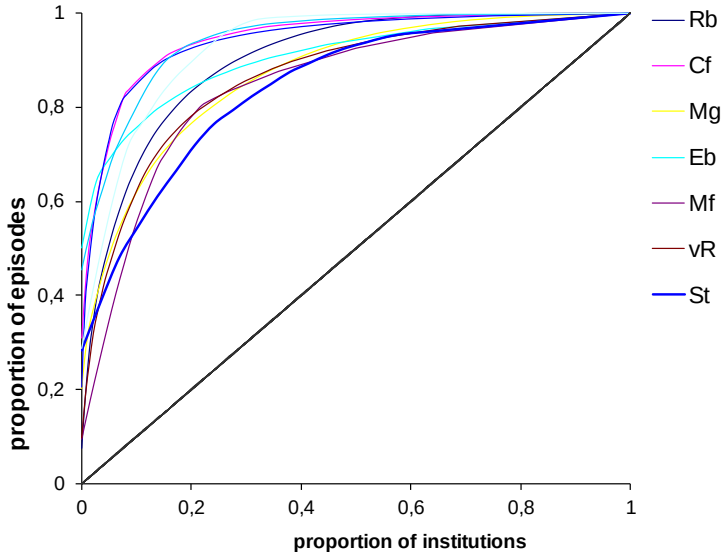
- Centralized care (50% of episodes in 25 centers)
- Available care (80% of patients living in 50 km from nearest center)



Centralized care without CE-designation



2. RD patient pathway analysis, HDR study (Hungary, 2004-2009)

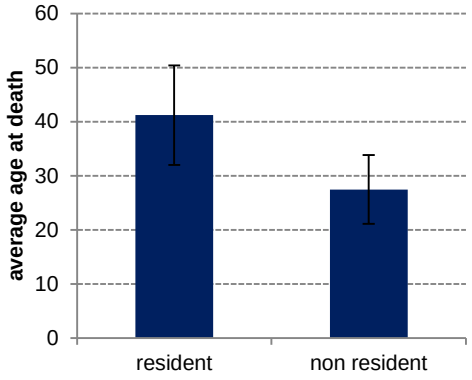


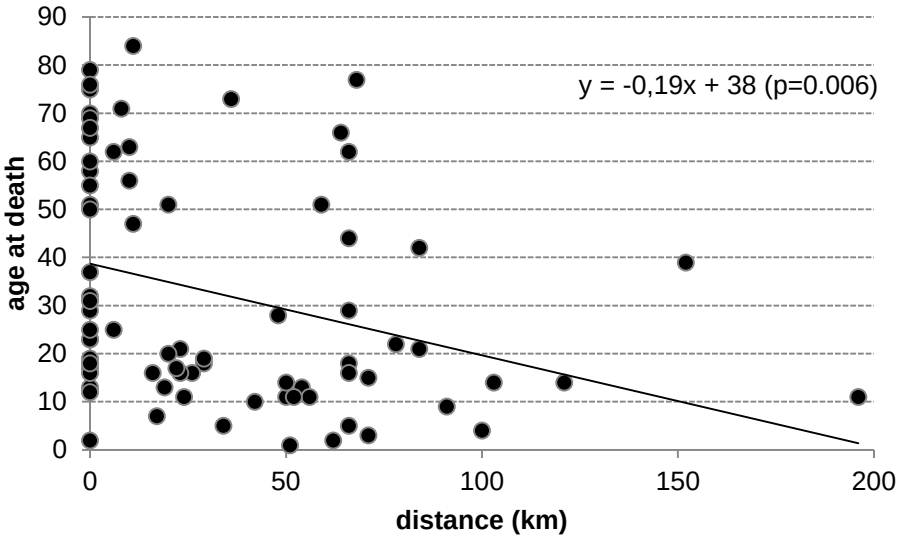
There are under-detected RDs: specific RD code does not work properly in case of underdiagnosing diseases.

	A	Episodes' 50%
tuberous sclerosis (underdetected)	0,34	4
epidermolysis bullosa (within reference range)	0,34	7
Marfan syndrome (underdetected)	0,36	10
von Recklinghausen syndrome (underdetected)	0,37	11
brain tumor (<18) (within reference range)	0,40	1
myasthaenia gravis (within reference range)	0,42	3
Down syndrome (within reference range)	0,44	4
cystic fibrosis (within reference range)	0,44	2
limphoid leukemia (<18) (within reference range)	0,44	3

2. Cystic fibrosis by discharge records

- The average **age at death** is higher among patients **living in towns with CF center** („residents”)
- The **distance** between residential place and CF center related to the age at death



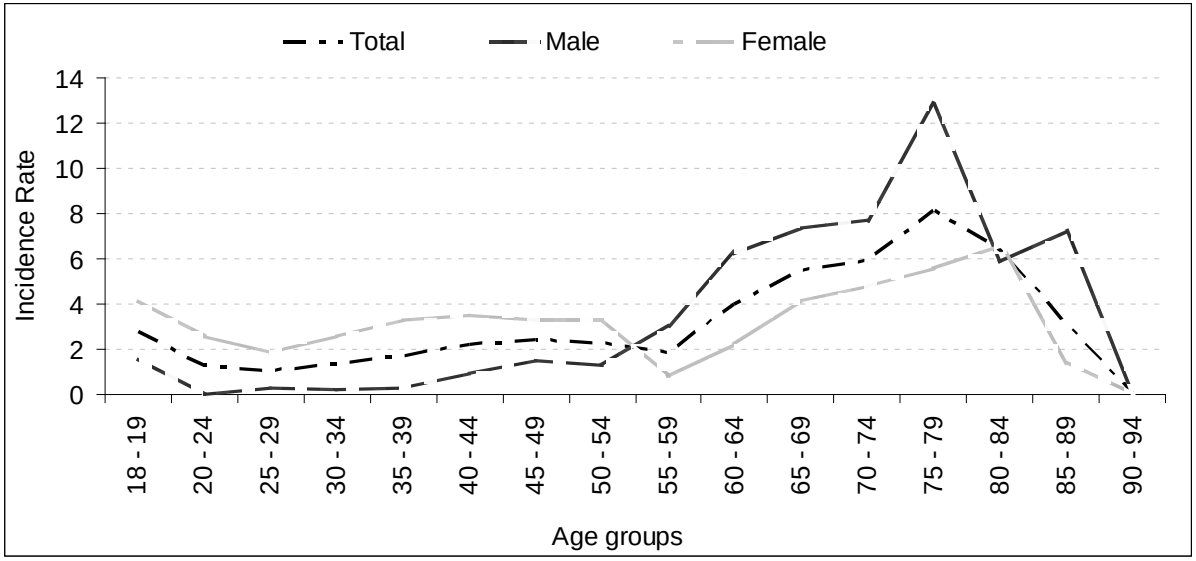


The age at death of patients dependent of residential place

2. Myasthenia gravis by discharge records

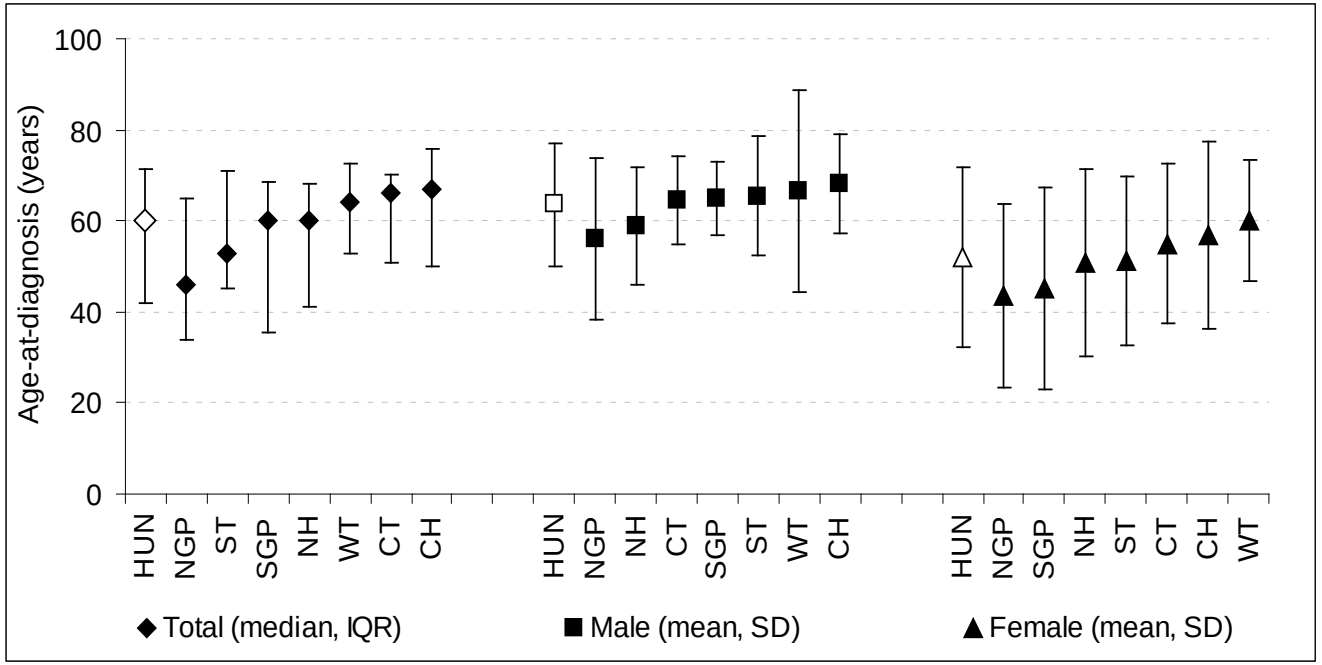
- The observed MG incidence rate: 2.76/100,000
- Reference range:
- 0.3-3/100,000

The incidence is in reference range

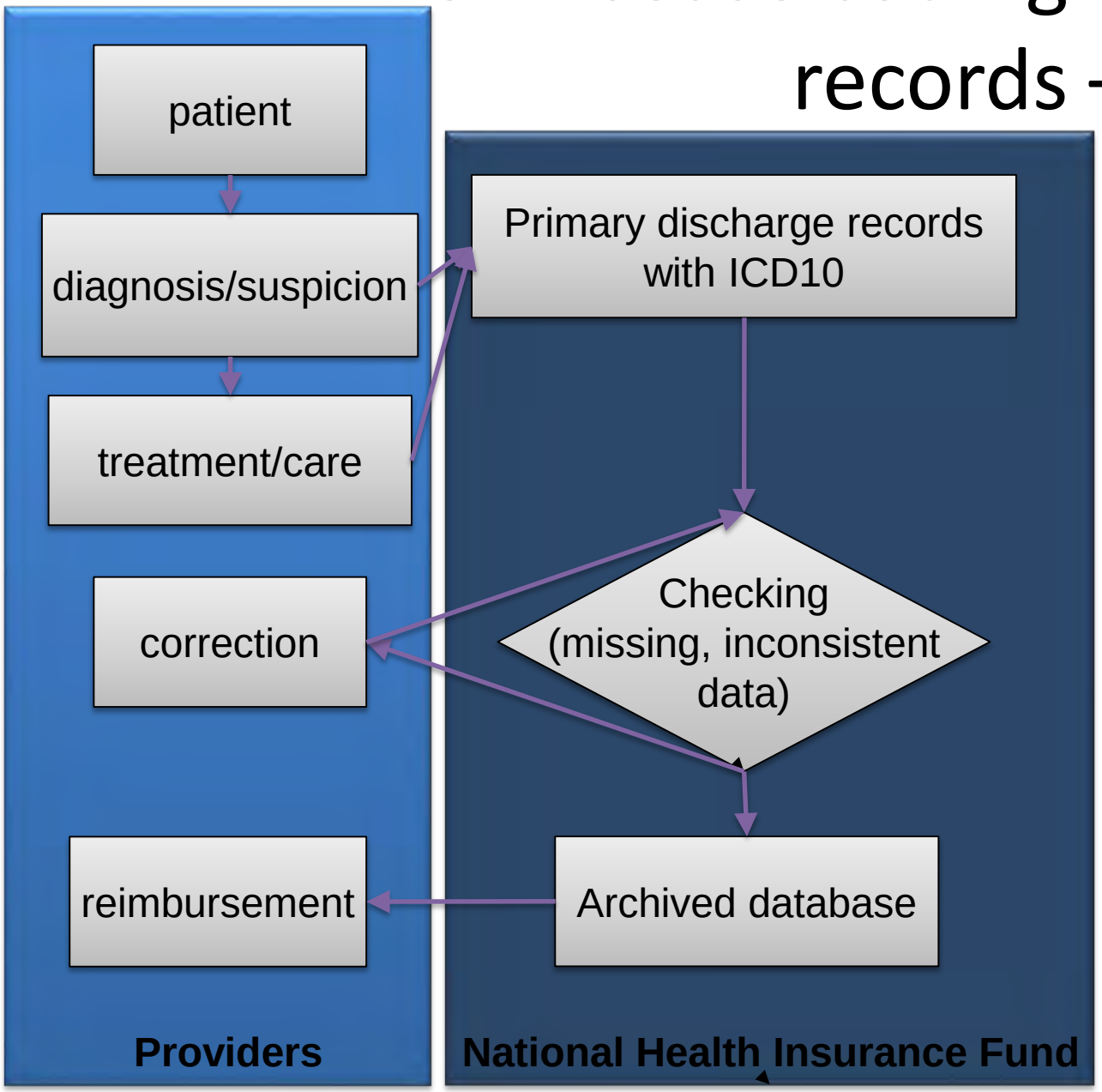


2. Myasthenia gravis by discharge records

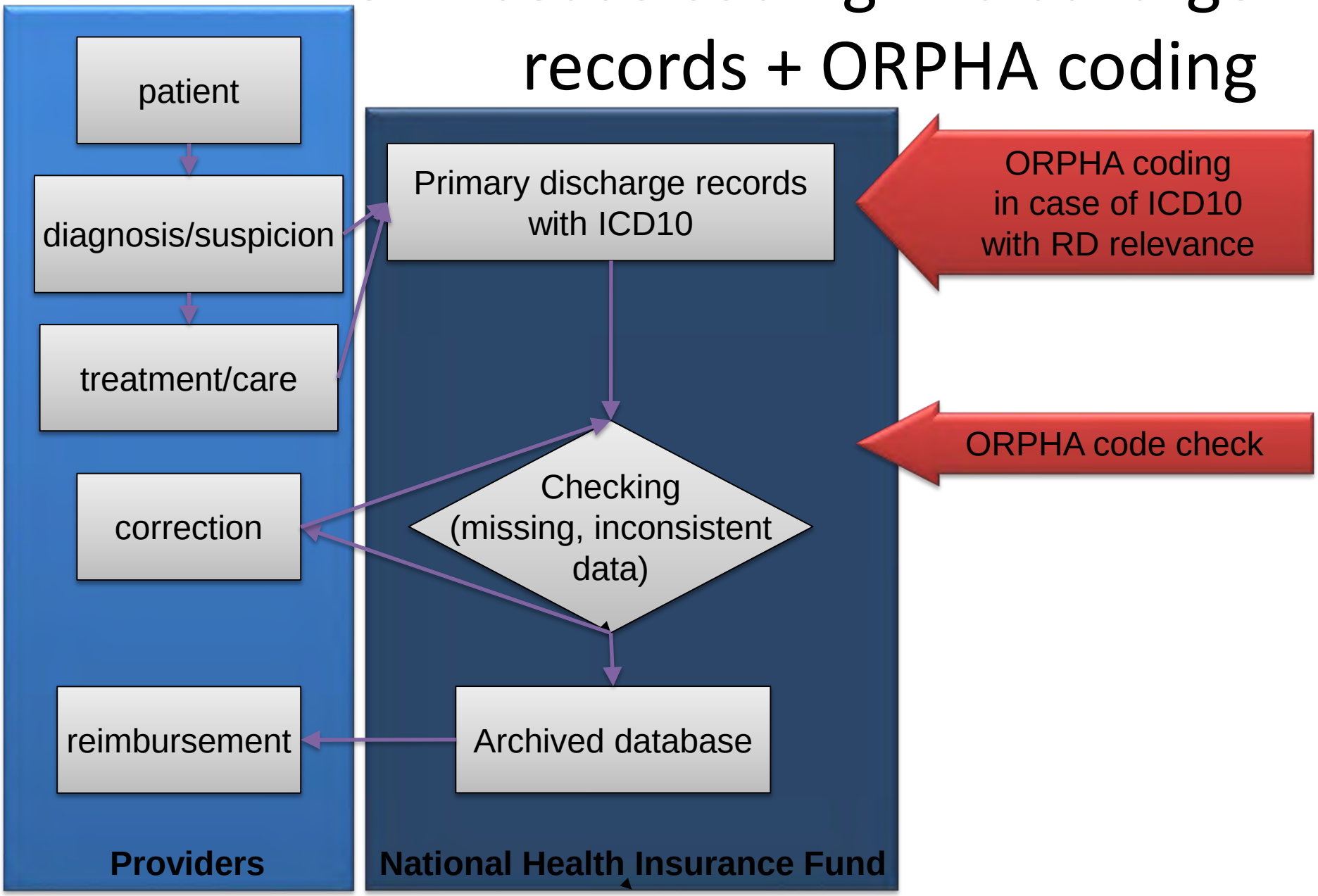
- The region specific average age-at-diagnosis showed spatial variability for both sexes (p=0.010)
- Age-at-diagnosis was not associated with the place of residence of patients for both males (p=0.091) and females (p=0.072)



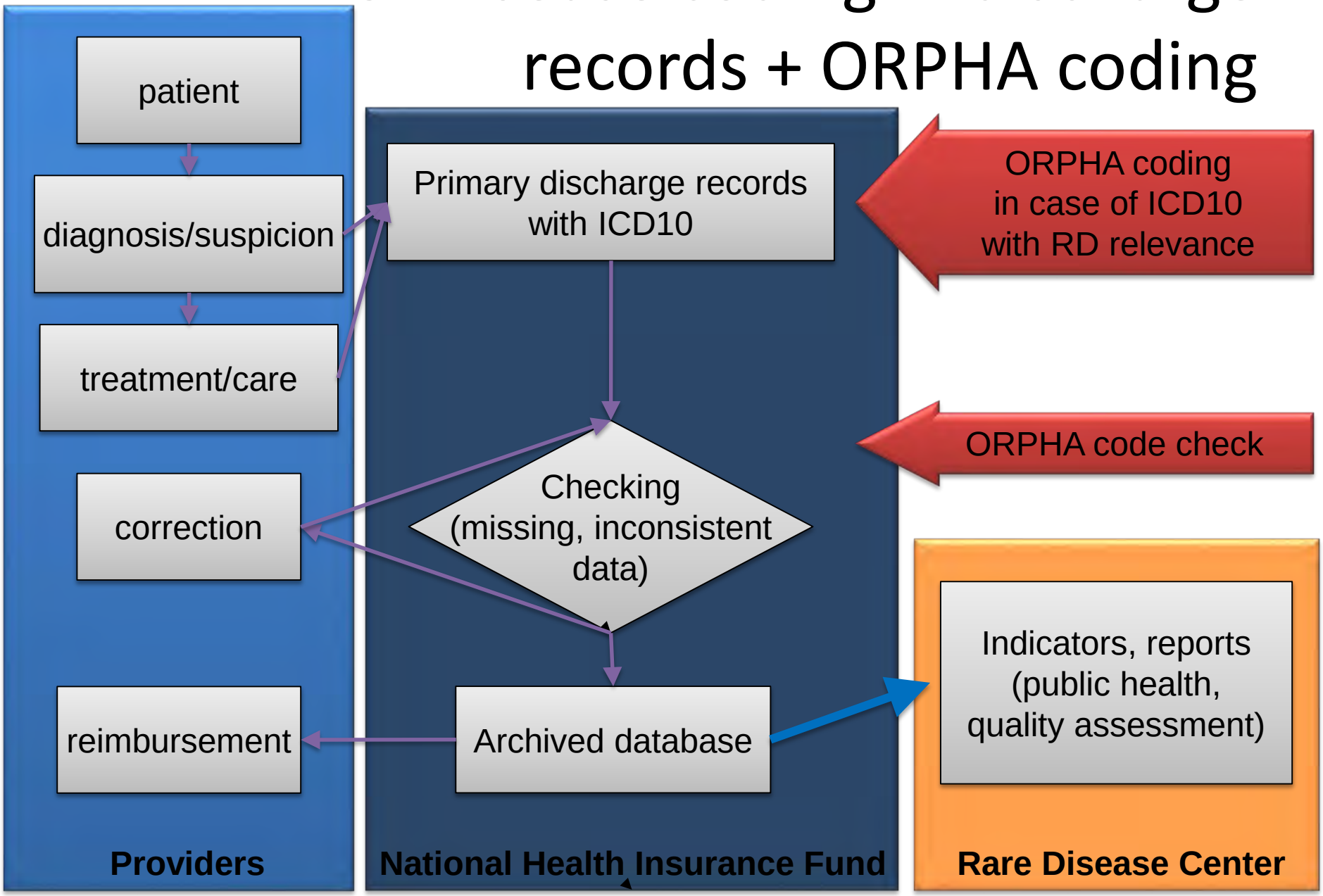
3. Disease coding in discharge records – at present



3. Disease coding in discharge records + ORPHA coding



3. Disease coding in discharge records + ORPHA coding



3. Teljesítmény-elszámolási jelentés + ORPHA kód

- BNO10 kód kiegészítve ORPHA kóddal befogadási feltétel
  - Ha több RD tartozik egy BNO kódhoz
  - Ha RD és gyakori betegség is tartozik a BNO kódhoz
- Minimális kódolási többletfeladat
- Nem igényel IT fejlesztést, jogszabály módosítást igényel, új jogi környezetet nem

3. Teljesítmény-elszámolási jelentés + ORPHA kód

- erőforrás:
  - ORPHA kód a 0/1/kevés esetszámra: a kódolás haszna minimális lehet esetleg nagy befektetés mellett
- kódolás technika:
  - ORPHA kód hierarchia (csoportnév→ RD): karakterszám
  - összetett kórképek kódolási szabálya
  - gyanú, ideiglenes diagnózis megkülönböztetése
- képzés:
  - az aluljelentés elkerülése érdekében
- Jogi befogadás:
  - Valakinek hivatalosan kell az ORPHA kódokat befogadni (és időnként felülvizsgálni)

ORPHA code for discharge records

- Orvosok, szakdolgozók:
  - Célokkal egyetértenek
  - Bizalmatlanság a teljesítmény-elszámolási rekordokkal szemben
- Döntéshozók:
  - Félelem a generált költségektől (többlet ellátástól)
  - Hiányzó ismeretek

- Fokozatos bevezetés
  - A monitoring demonstrált haszna a legjobb érv

Folytatás

- Mortalitási és OEP/NEAK adatbázisok feldolgozása (521 specifikus kód, 167 csoportkód, 8 tőr és csillag; sok alulregisztrált)
- Regiszterek építése
- ICD11 bevezetés
- DR kiegészítés – EESZT

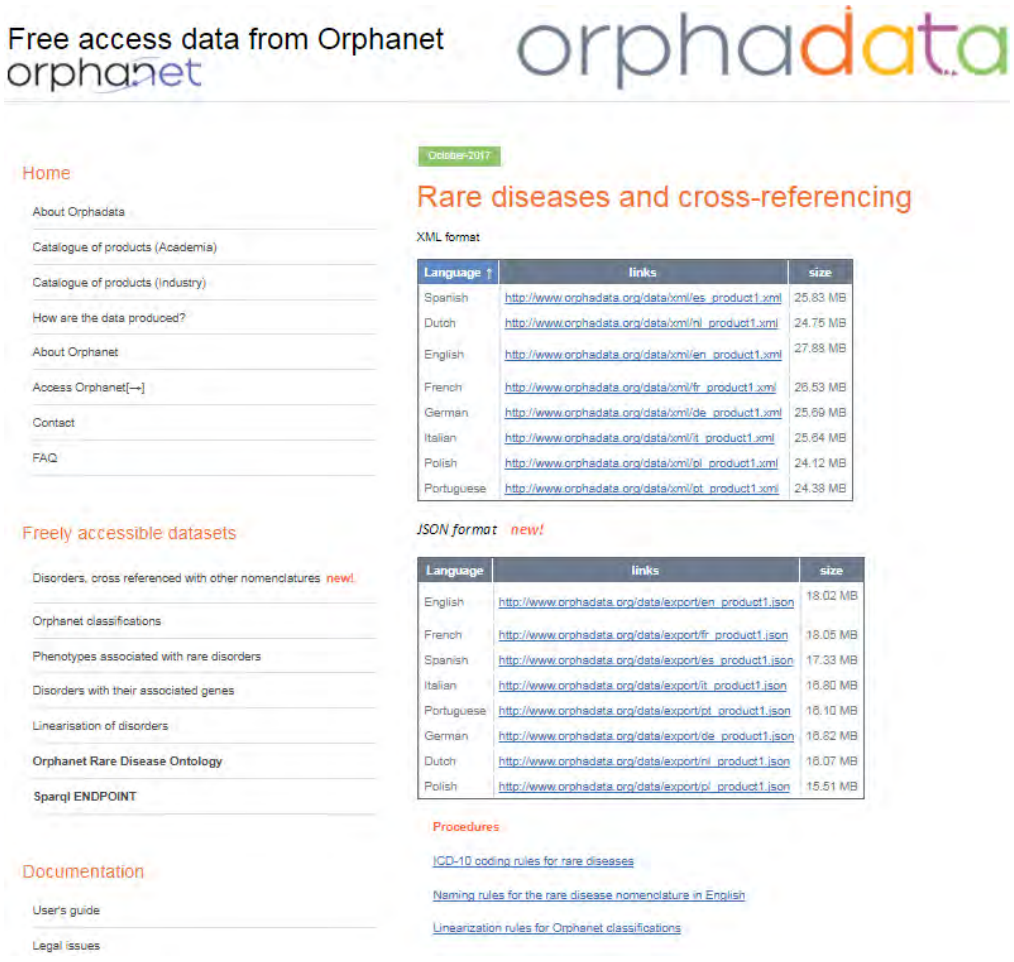
Common data elements (JRC)

GROUP	ELEMENT N°	ELEMENT NAME	ELEMENT DESCRIPTION	CODING	COMMENT
1. Pseudonym	1.1.	Pseudonym	Patient's pseudonym	• String	The JRC is working on providing a pseudonymisation tool to the registries
2. Personal information	2.1.	Date of birth	Patient's date of birth	• Date (dd/mm/yyyy)	
	2.2.	Sex	Patient's sex at birth	• Female • Male • Undetermined • Foetus (Unknown)	
3. Patient Status	3.1.	Patient's status	Patient alive or dead	• Alive • Dead • Lost in follow-up • Opted-out	If dead then answer question 3.2
	3.2.	Date of death	Patient's date of death	• Date (dd/mm/yyyy)	
4. Care pathway	4.1.	First contact with specialised centre	Date of first contact with specialised centre	• Date (dd/mm/yyyy)	

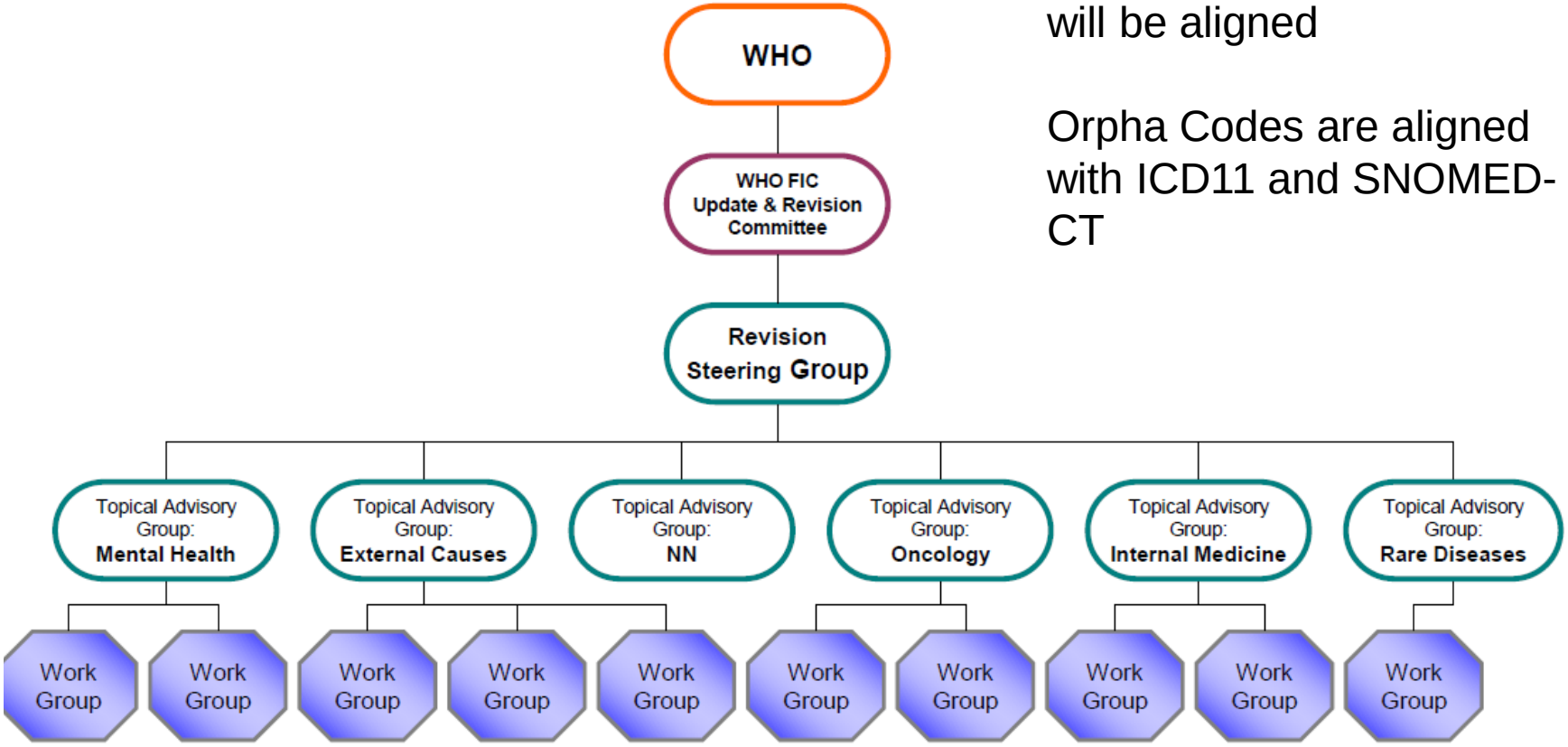
Common data elements (JRC)

5. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
	5.2.	Age at diagnosis	Age at which diagnosis was made	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
6 Diagnosis	6.1.	Diagnosis of the rare disease	Diagnosis retained by the specialised centre	Orpha code (strongly recommended – see link) / Alpha code/ ICD-9 code/ ICD-9-CM code / ICD-10 code	http://www.orphadata.org/cgi-bin/inc/product1.inc.php
	6.2.	Genetic diagnosis	Genetic diagnosis retained by the specialised centre	International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code	http://www.hgvs.org
	6.3	Undiagnosed case	How the undiagnosed case is defined	• Phenotype (HPO) • Genotype (HGVS)	
7. Research	7.1.	Agreement to be contacted for research purposes	Patient's permission exists for being contacted for research purposes	• YES • NO	
	7.2.	Consent to the reuse of data	Patient's consent exists for his/her data to be reused for other research purposes	• YES • NO	
	7.3.	Biological sample	Patient's biological sample available for research	• YES • NO	If YES answer question 7.4
	7.4.	Link to a biobank	Biological sample stored in a biobank	• YES (if appropriate use link) • NO	https://directory.bbmri-eric.eu
8.Disability	8.1.	Classification of functioning/disability	Patient's disability profile according to International Classification of Functioning and Disability (ICF)	• Disability profile / Score	http://www.who.int/classifications/icf/whodasii/en/

Common data elements (JRC)



ICD-11 (2018)



ICD11 and SNOMED-CT will be aligned

Orpha Codes are aligned with ICD11 and SNOMED-CT

ICD-11 (content model)

1. **ICD Concept Title**
   - 1.1. Fully Specified Name

2. **Classification Properties**
   - 2.1. Parents
   - 2.2 Type
   - 2.3. Use and Linearization(s)

3. **Textual Definition(s)**

4. **Terms**
   - 4.1. Base Index Terms
   - 4.2. Inclusion Terms
   - 4.3. Exclusions

5. **Body Structure Description**
   - 5.1. Body System(s)
   - 5.2. Body Part(s) [Anatomical Site(s)]
   - 5.3. Morphological Properties

6. **Manifestation Properties**
   - 6.1. Signs & Symptoms
   - 6.2. Investigation findings

7. **Causal Properties**
   - 7.1. Etiology Type
   - 7.2. Causal Properties - Agents
   - 7.3. Causal Properties - Causal Mechanisms
   - 7.4. Genomic Linkages
   - 7.5. Risk Factors

8. **Temporal Properties**
   - 8.1. Age of Occurrence & Occurrence Frequency
   - 8.2. Development Course/Stage

9. **Severity of Subtypes Properties**

10. **Functioning Properties**
    - 10.1. Impact on Activities and Participation
    - 10.2. Contextual factors
    - 10.3. Body functions

11. **Specific Condition Properties**
    - 11.1 Biological Sex
    - 11.2. Life-Cycle Properties

12. **Treatment Properties**

13. **Diagnostic Criteria**

RD-ACTION

• There are very few implemented strategies to produce statistics on rare diseases at national level. Nevertheless, the production of statistics on rare diseases is among the strategies included in many national plans/strategies, most of them being "in design" at the moment.

• RD patients are mostly coded in health information systems in the framework of the use of general coding system for morbidity/mortality, namely ICD-10.

• Multiple terminologies are used for coding diseases and for phenotypes.

• Orphacodes emerge as the main coding system dedicated to RD in both inpatient and outpatient settings.

• In the majority of MS answering the survey (n=15) a unique and stable patient identifier is in use. This is an important prerequisite for evaluating RD impact at population level and on the health care systems if orphacodes are adopted, as it allows the tracing of RD patients across different care settings and thus health information systems

• The RD coding evolving and articulated situation at EU level is a strong base for the adoption and implementation of D5.2 guidelines: Each of the 6 guidelines are up to at least 60% "planned" or "ongoing" across MS, and 12% were deemed "done" at the time of this survey.

R-kód (Deák György)

Kórkép	ORPH	R-kó	J	Prevalenc	nherita	ge of or	ICD-	ICD-	név2	név3	név4	név5	név6	név7
10p11.21p12.31 microdeletion syndrome	284169	R06082		<1 / 1 000 0(	Sporadic	Neonatal/	Q83.5		10p12p11 microd	Deletion 10p11.2:	Monosomy 10p1:	del(10)(p11.21p12.31)		
10q22.3q23.3 microdeletion syndrome	276413	R05987		Unknown	Autosom	Neonatal/	Q93.5		Del(10)(q22.3q23.	Deletion 10q22.3(	Monosomy 10q22.3q23.3			
10q22.3q23.3 microduplication syndrome	276422	R05988		Unknown	Autosom	Neonatal/	Q92.3		Dup(10)(q22.3q2:	Trisomy 10q22.3q23.3				
11p15.4 microduplication syndrome	300305	R06338		<1 / 1 000 0(	Autosom	Neonatal/	Q92.3		Dup(11)p(15.4)	Trisomy 11p15.4				
12p12.1 microdeletion syndrome	313884	R06468		<1 / 1 000 0(	Autosom	Neonatal/	Q93.5		Del(12)(p12.1)	Monosomy 12p12.1				
12q14 microdeletion syndrome	94063	R03837	S	<1 / 1 000 0(	Unknown	Childhooc	Q93.5		Del(12)(q14)	Deletion 12q14	Monosomy 12q1	Osteopoikilosis - short stature - intellectual deficit		
12q15q21.1 microdeletion syndrome	289513	R06130		-	-	Neonatal/	Q93.5		Del(12)(q15)(q21.	Deletion 12q15q2	Monosomy 12q15q21.1			
14q11.2 microdeletion syndrome	261120	R05799	S	<1 / 1 000 0(	Sporadic	Neonatal/	Q93.5		Del(14)(q11.2)	Monosomy 14q11.2				
14q11.2 microduplication syndrome	261229	R05807		<1 / 1 000 0(	-	-	Q92.3		Dup(14)(q11.2)	Trisomy 14q11.2				
14q12 microdeletion syndrome	261144	R05800	S	<1 / 1 000 0(	-	Neonatal/	Q93.5		Del(14)(q12)	Monosomy 14q12				
14q22q23 microdeletion syndrome	264200	R05879		<1 / 1 000 0(	-	-	Q93.5		14q22-q23 micro(	Del(14)(q22q23)	Monosomy 14q2:	Monosomy 14q22q23		
15q overgrowth syndrome	314585	R06501		<1 / 1 000 0(	Sporadic	Neonatal/	Q87.3							
15q11.2 microdeletion syndrome	261183	R05801		<1 / 1 000 0(	-	-	Q93.5		Del(15)(q11.2)	Monosomy 15q11.2				
15q11q13 microduplication syndrome	238446	R05541	S	<1 / 1 000 0(	Sporadic	Neonatal/	Q92.3		15q11-q13 duplic	15q11-q13 micro(	15q11q13 duplica	Dup(15)(q11q13)	Trisomy 15q11-q1	Trisomy 15q11q13
15q13.3 microdeletion syndrome	199318	R05194	S	Unknown	Autosom	Childhooc	Q93.5		Del(15)(q13.3)	Monosomy 15q13.3				
15q14 microdeletion syndrome	261190	R05802	S	<1 / 1 000 0(	Sporadic	Neonatal/	Q93.5		Del(15)(q14)	Monosomy 15q14				

6782 definíció

Does a national strategy to produce statistics on rare diseases exist in your country?