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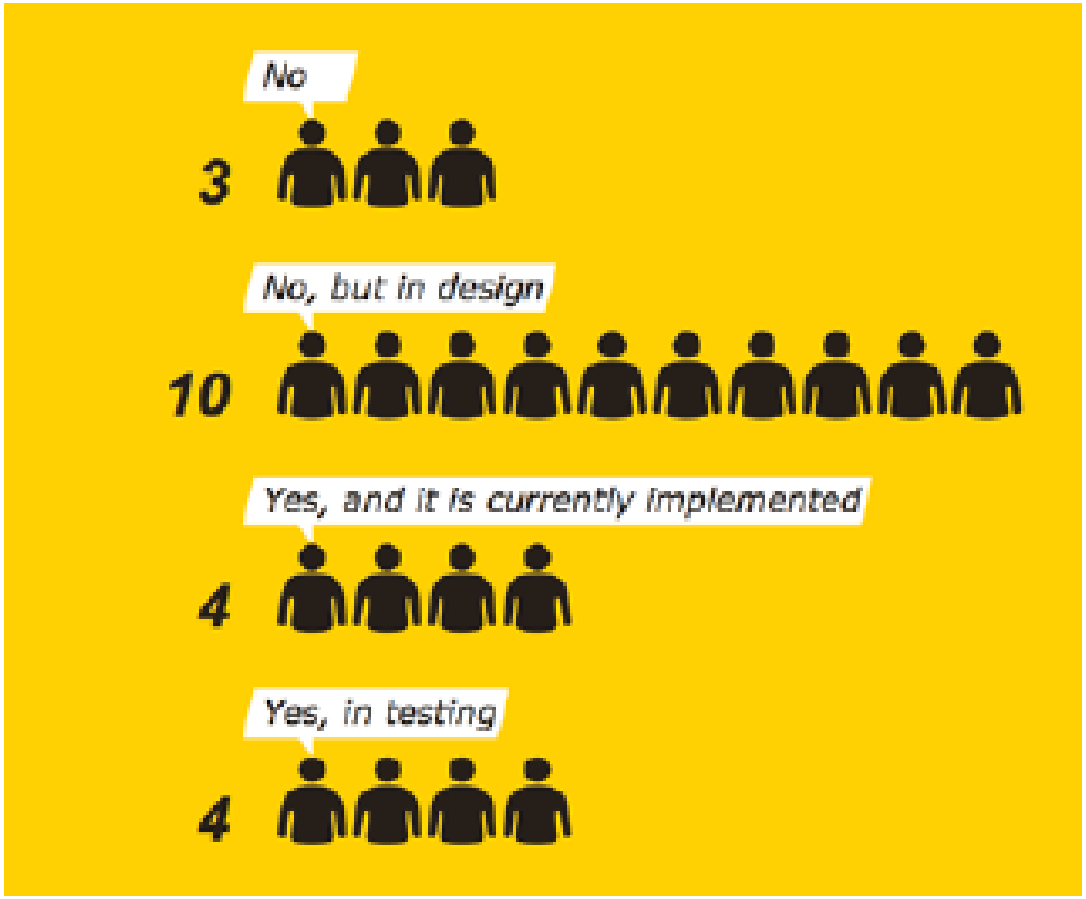
Indicators on the epidemiology of rare diseases with independent ICD codes: Hungarian experiences

Conference

for an EU commitment to tackling rare diseases

2024, Budapest

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



RD-ACTION, 2015

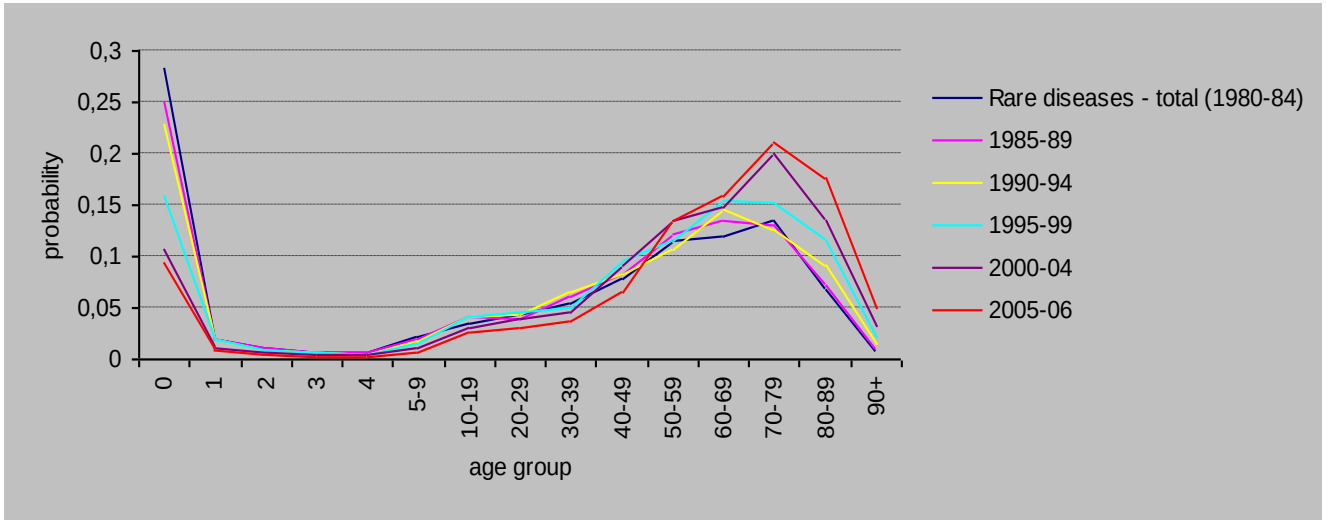
The European Health Data Space for complex systems management

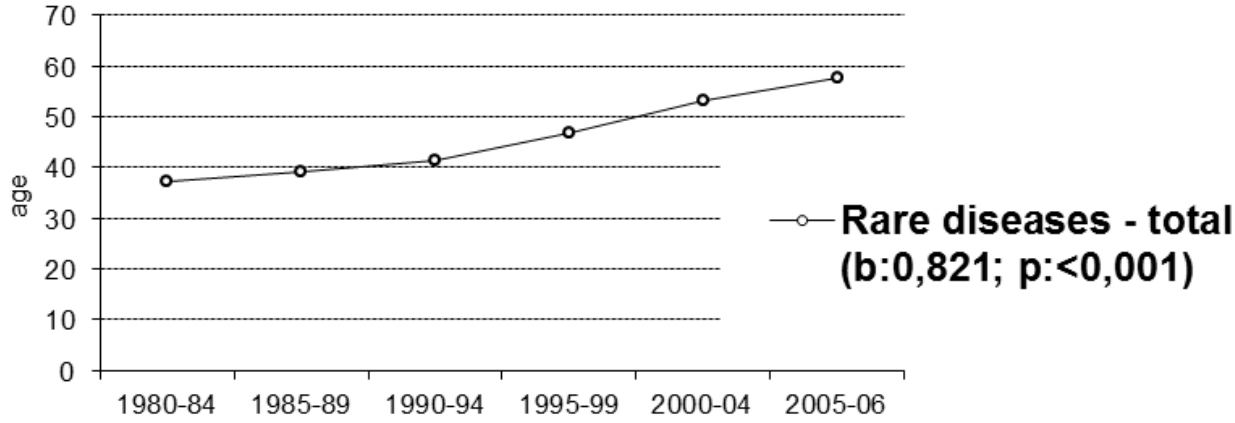
- Set common and measurable goals that will help define and implement national plans and strategies for RD:
  1. **improved health outcomes**
  2. **reduced inequality**
  3. **boosting innovation**
- **General solution is Orpha-coding, but till?**
  - 696 distinct rare diseases **by ICD-10 codes** (Orphanet, Ana Rath)
  - Available **datasets with ICD-10 codes covering the whole population**:
    1. cause of death data (Central Statistical Office)
    2. discharge records of secondary care (National Health Insurance Fund)

Cause of death

Mean age-at-death

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





Cause of death

Indicators derived from the mean age-at-death

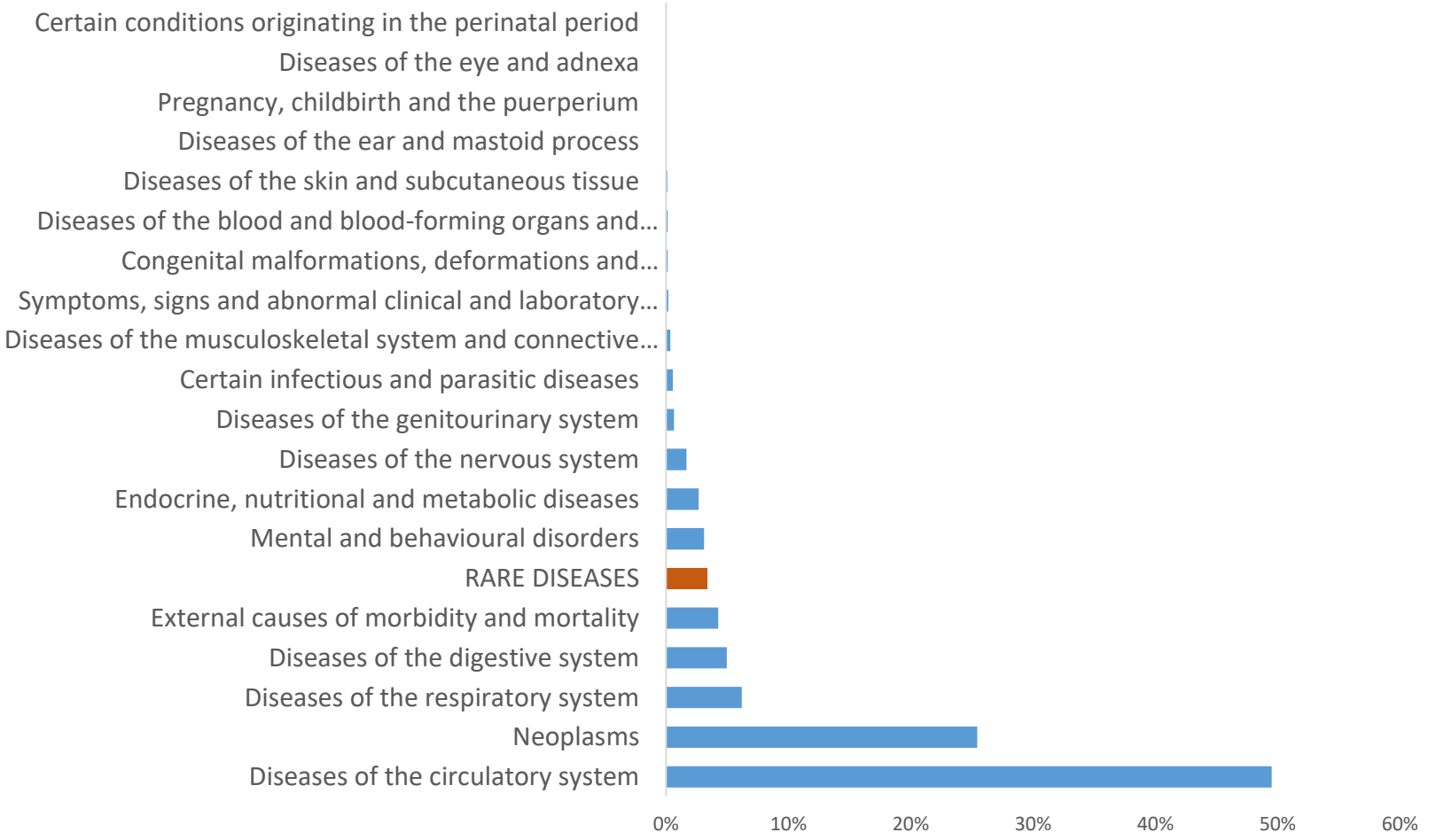
Rare diseases mortality, Hungary, (1980-2006; N=44775)

Indicators	1980-1984	2005-2006
Mean age at death attributed to rare diseases (p for change < 0.001)	**37.3**	**57.7**
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]**
Potential years of life lost attributed to rare diseases (PYLL)	**66119**	**21900**

Cause of death

Relative importance of rare disease mortality

(2006-2017, adults, Hungary, N=4422/year, 3.4%)



Cause of death

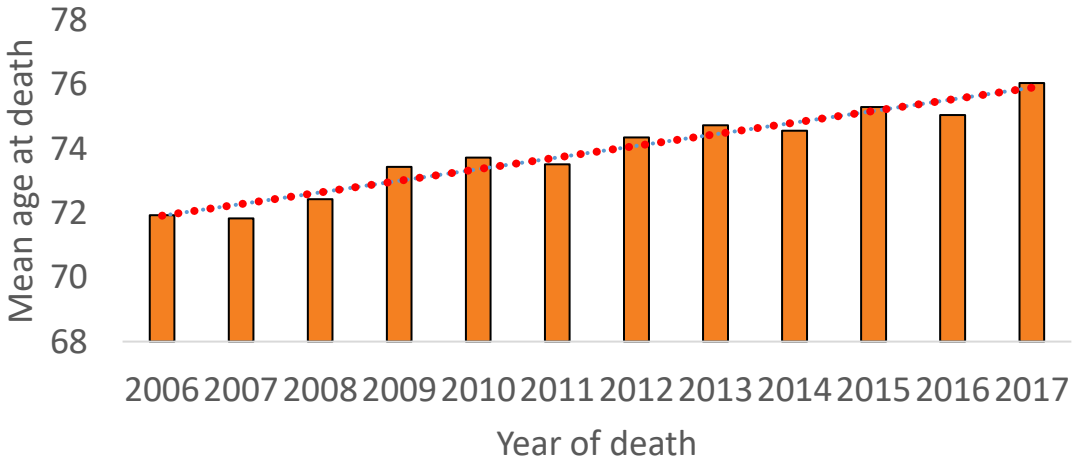
Change of mean age-at-death

(2006-2017, adults, Hungary)

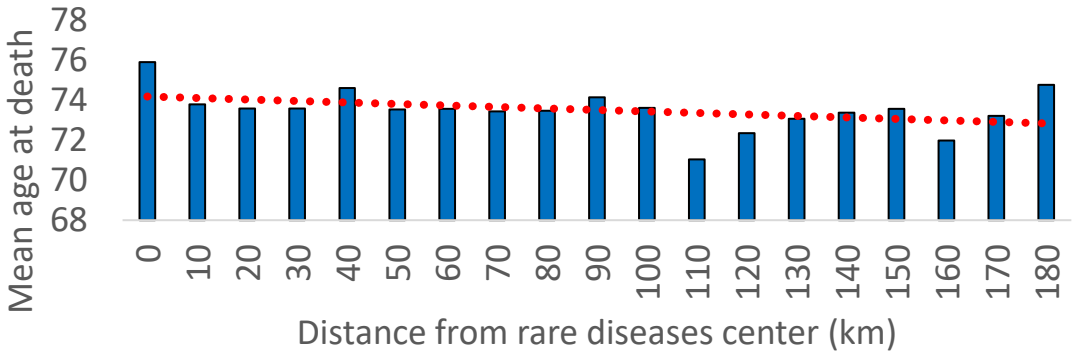
	time (year)	sex (females/males	distance (10 km)
Cancer	0.254 [0.185;0.323]	3.563 [3.092;4.034]	-0.007 [-0.012;-0.002]
Metabolic	0.222 [-0.08;0.523]	8.858 [6.728;10.98]	-0.015 [-0.038;0.009]
Mental	0.174 [0.143;0.205]	3.425 [3.202;3.649]	-0.018 [-0.02;-0.015]
Neurologic	0.421 [0.22;0.621]	5.27 [3.895;6.644]	-0.026 [-0.041;-0.011]
Cardiovascular	0.16 [0.088;0.232]	6.445 [5.915;6.974]	-0.012 [-0.018;-0.006]
Gastrointestinal	0.078 [-0.167;0.322]	5.733 [3.964;7.502]	-0.017 [-0.037;0.002]
Musculoskeletal	0.185 [-0.023;0.394]	3.097 [1.323;4.871]	-0.023 [-0.038;-0.007]
Congenital anomalies	0.493 [0.233;0.752]	2.673 [0.881;4.465]	0 [-0.019;0.02]
Rare diseases	**0.355 [0.32;0.39]**	**7.830 [7.59;8.069]**	**-0.016 [-0.019;-0.013]**

Cause of death

Change of mean at death by year and distance from the center of expertise



+0.36 year increase of the mean age at death per year

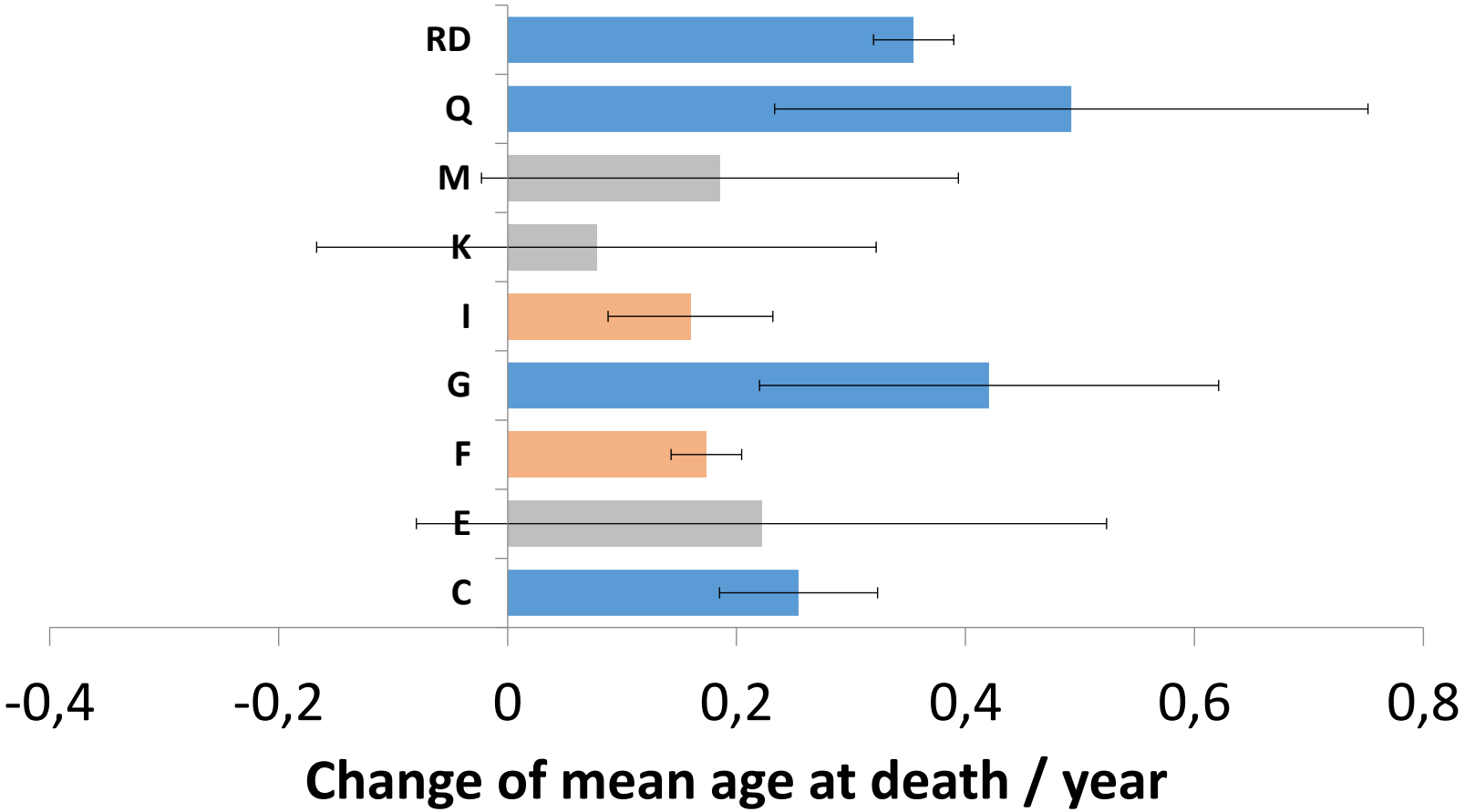


-0.75 year decrease of the mean age at death per 10 km

Cause of death

Change of mean age at death/year by ICD

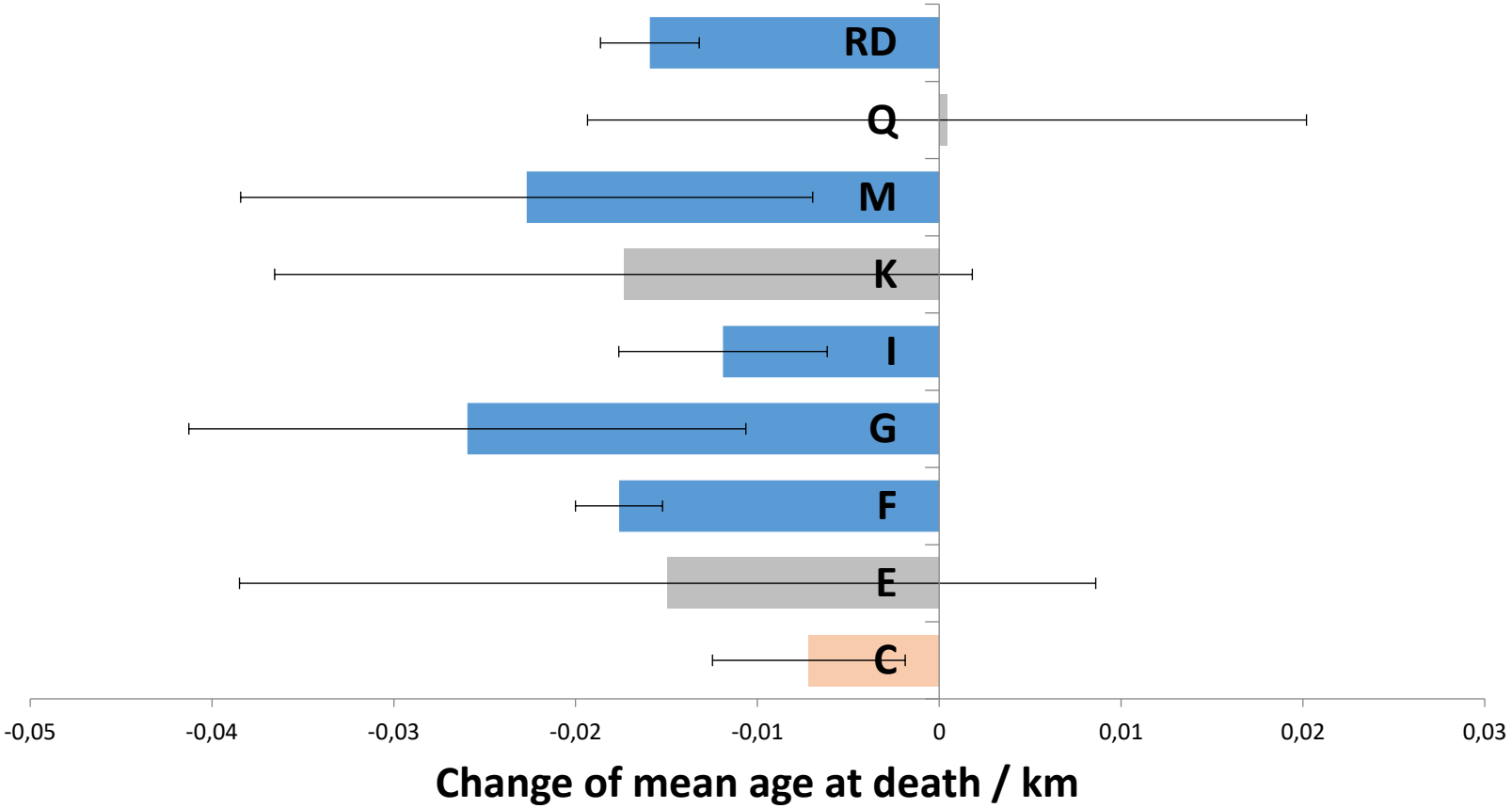
(2006-2017, Hungary)



Cause of death

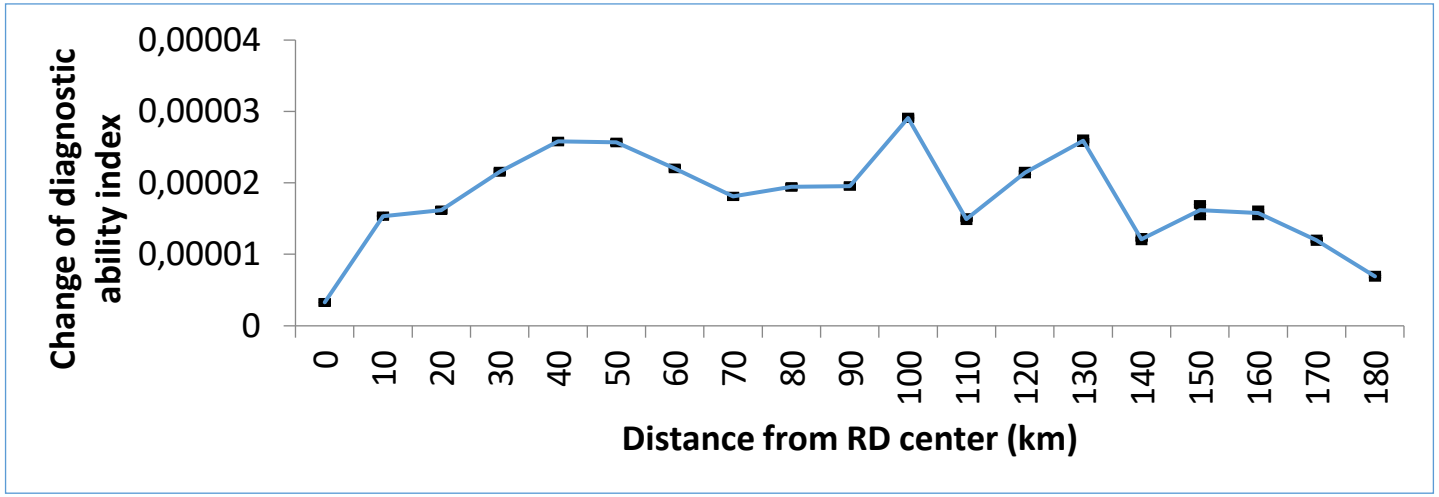
Change of mean at death/distance from the center of expertise by ICD

(2006-2017), Hungary

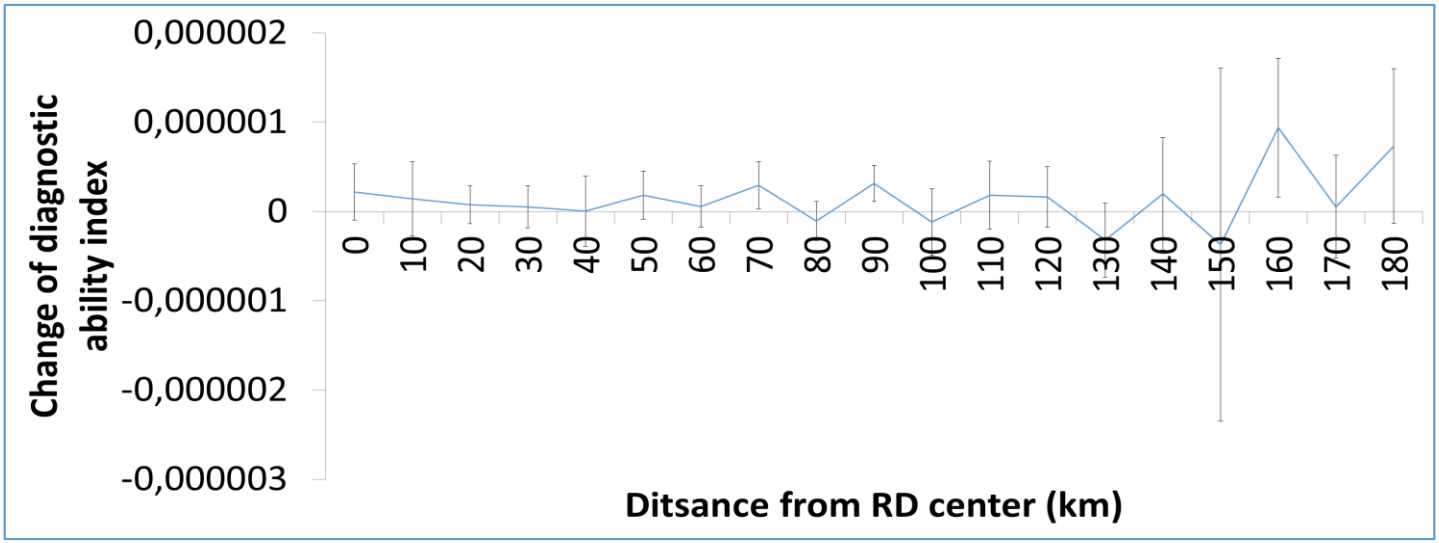


Cause of death

Diagnostic ability index (probability of RD in the population)



RD with distinct ICD10 codes



Myasthenia gravis

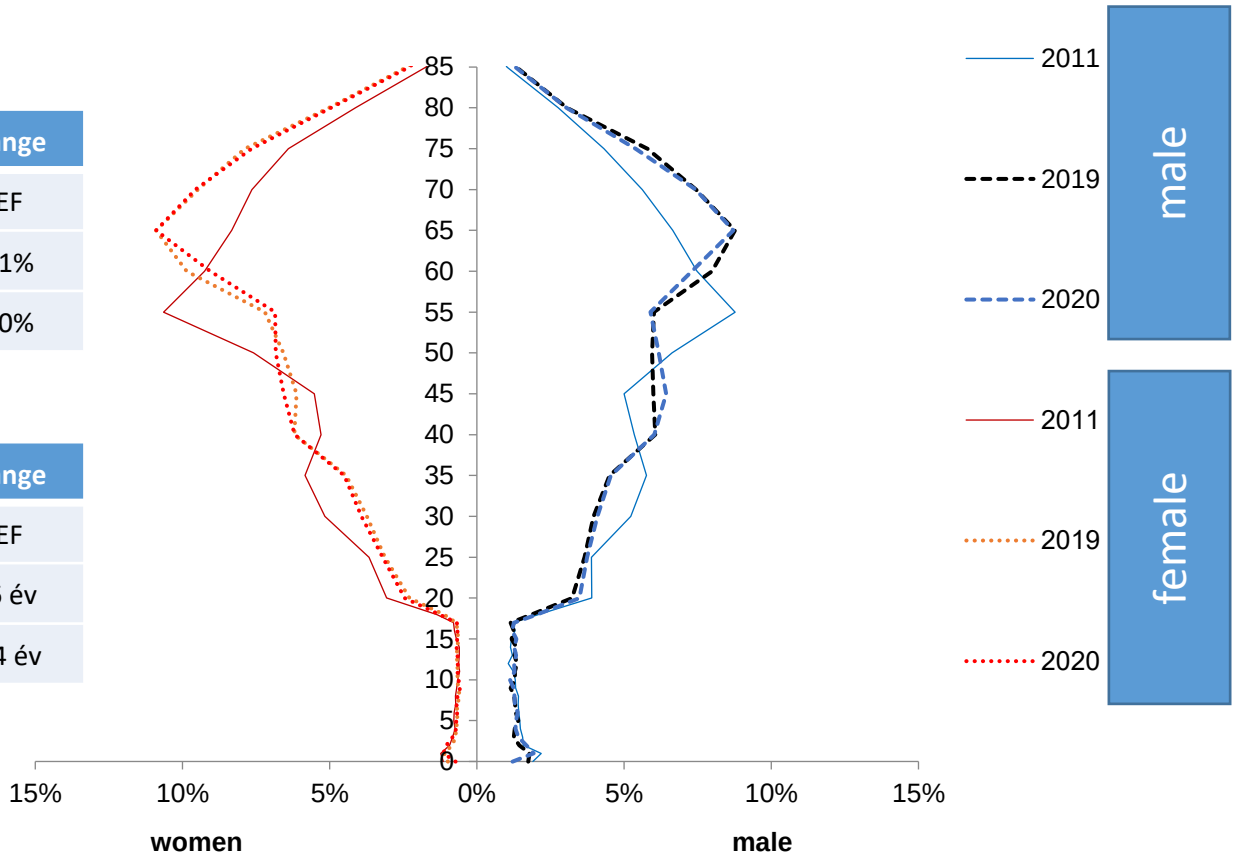
Discharge records

Prevalence of rare diseases

Discharge records of secondary care (National Health Insurance Fund)

	Cases	Change
2011	154 641	REF
2019	171 863	111%
2020	200 562	130%

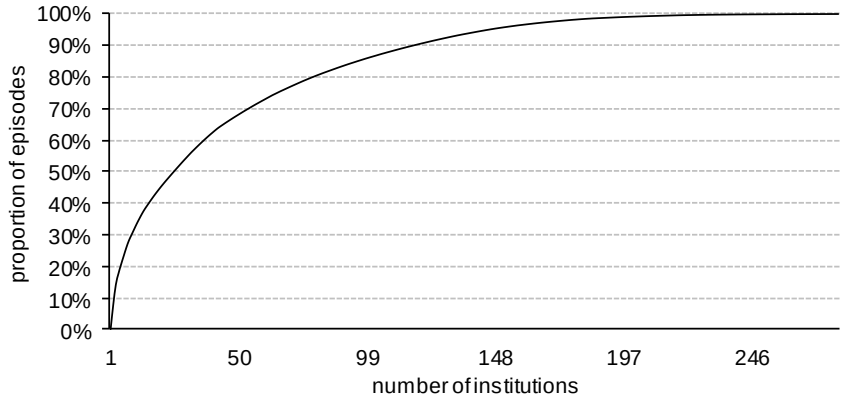
	Mean age	Change
2011	46.0 y	REF
2019	48.7 y	2,6 év
2020	48.3 y	-0,4 év



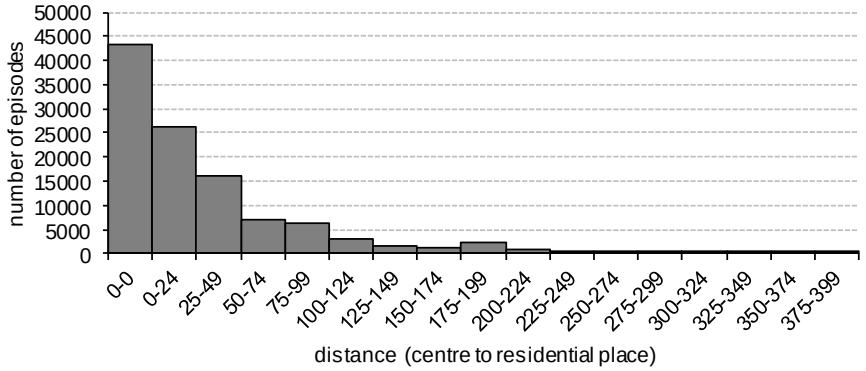
Discharge records

RD patient pathway indicators
(Hungary, 2004-2009)

- Number of centers providing care for 50% of episodes (25 centers, centralization of the care)



- Proportion of patients living in 50 km vicinity of the nearest center (80% of patients; geographical availability of the care)



Discharge records

Visiting GP a year

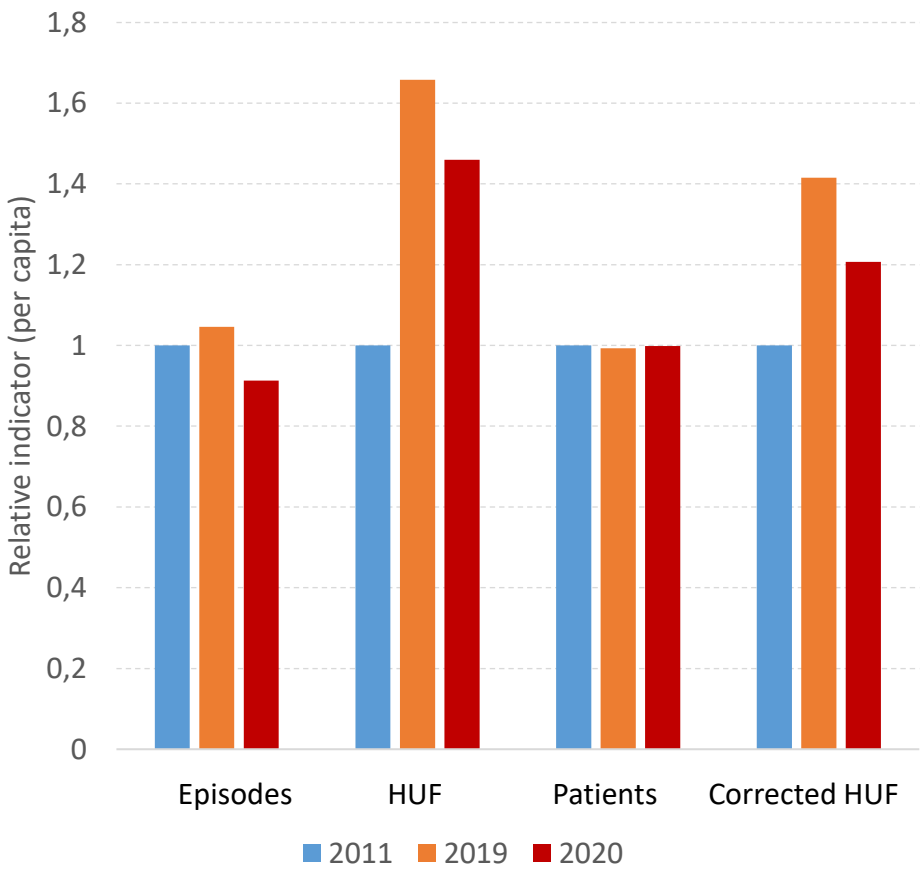
	Number of cases	Number of visits by a patient a year	Change
2011	2 003 806	13.0	REF
2019	2 212 595	12.9	99%
2020	2 449 851	12.2	94%

Discharge records

Use of outpatient centers' services

	Number of cases	Number of episodes	Billion HUF	Corrected billion HUF
2011	150 828	1 242 411	3.1	3.1
2019	166 472	1 443 989	5.7	5.0
2020	195 343	1 470 913	5.9	5.0

	Number of cases / registered cases	Number of episodes / registered cases	HUF / registered cases	Corrected HUF / registered cases
2011	0.98	8.03	20 050	20 050
2019	0.97	8.40	33 239	29 345
2020	0.97	7.33	29 271	24 992

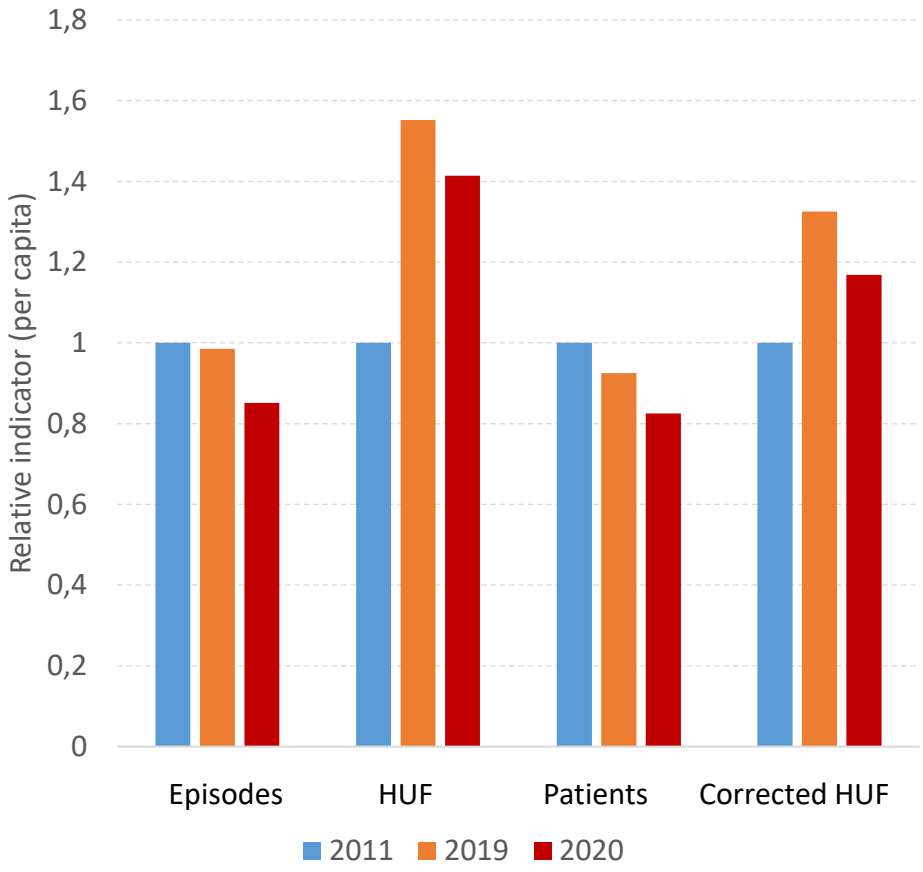


Discharge records

Use of inpatient services

	Number of cases	Number of episodes	Billion HUF	Corrected billion HUF
2011	39 495	95 686	20	20
2019	40 592	104 779	34	30
2020	42 278	105 634	37	31

	Number of cases / registered cases	Number of episodes / registered cases	HUF / registered cases	Corrected HUF / registered cases
2011	0.26	0.62	128 941	128 941
2019	0.24	0.61	200 110	176 665
2020	0.21	0.53	182 282	155 634



Discharge records

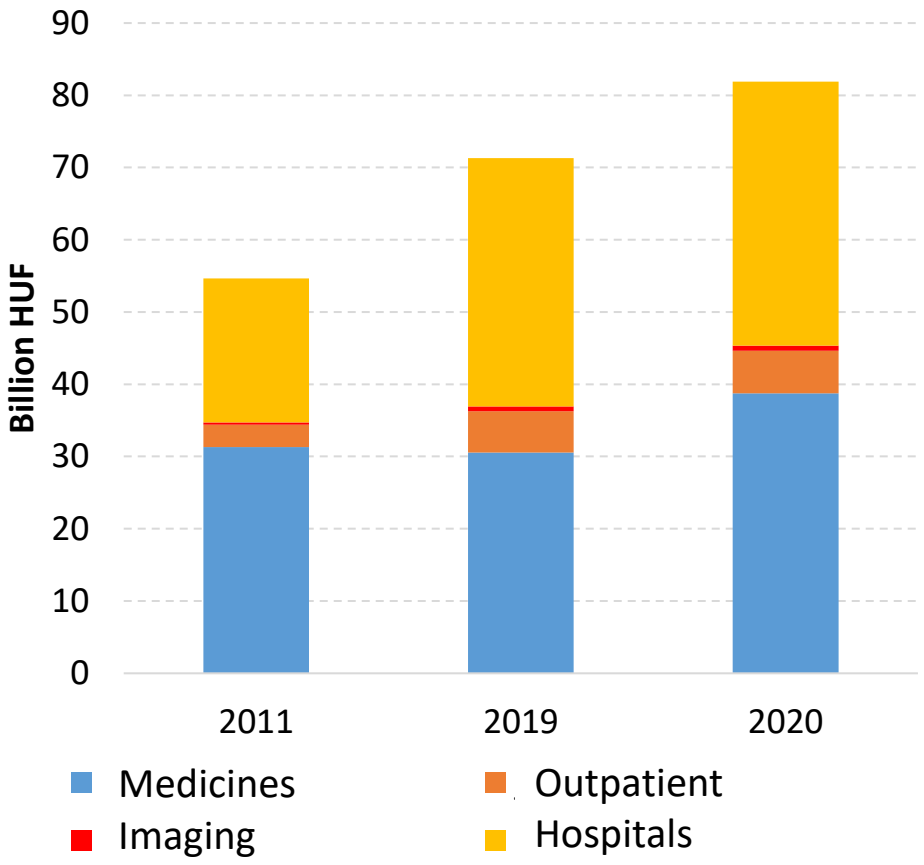
Health insurance fund payments for medications

	Billion HUF	Thousand HUF/patient	Change	Corrected billion HUF	Corrected thousand HUF/patient	Change
2011	31.3	202.4	REF	31.3	202.4	REF
2019	30.6	177.8	88%	27.0	157.0	78%
2020	38.8	193.2	95%	33.1	165.0	82%

Discharge records

Health insurance fund payments (billion HUF a year)

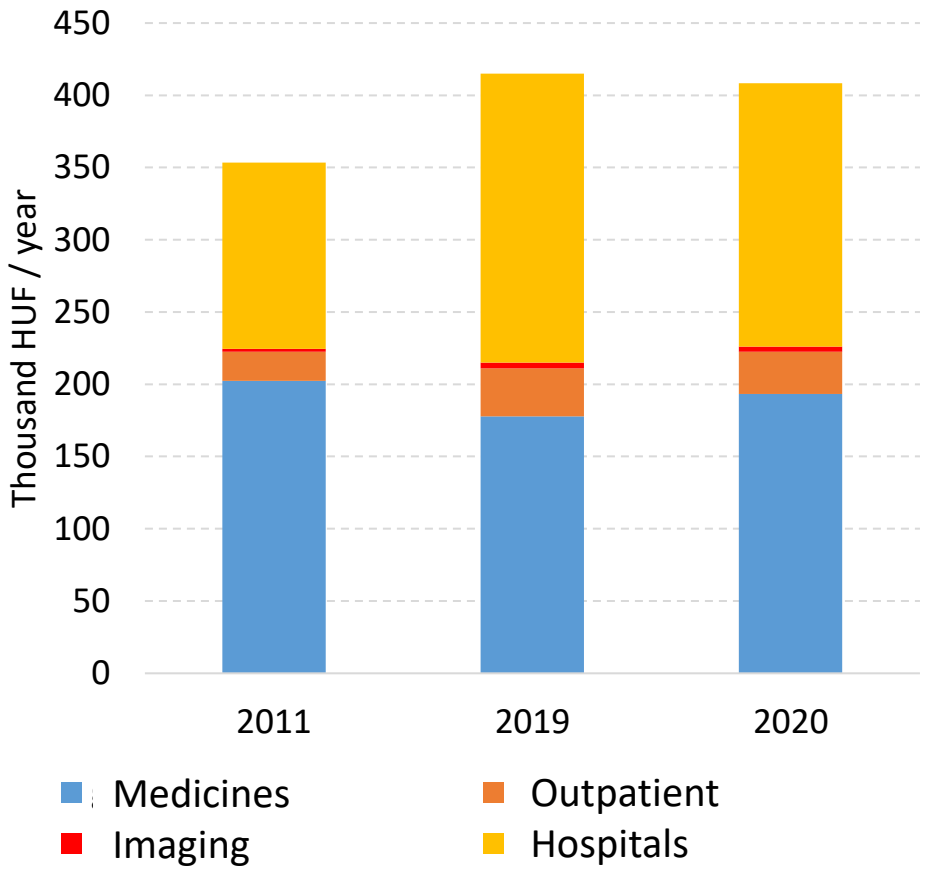
	Medicines	Outpatient services	CT/MRI imaging	Hospitals
2011	31.3	3.1	0.3	19.9
2019	30.6	5.7	0.7	34.4
2020	38.8	5.9	0.7	36.6



Discharge records

Health insurance fund payments per capita (thousand HUF a year)

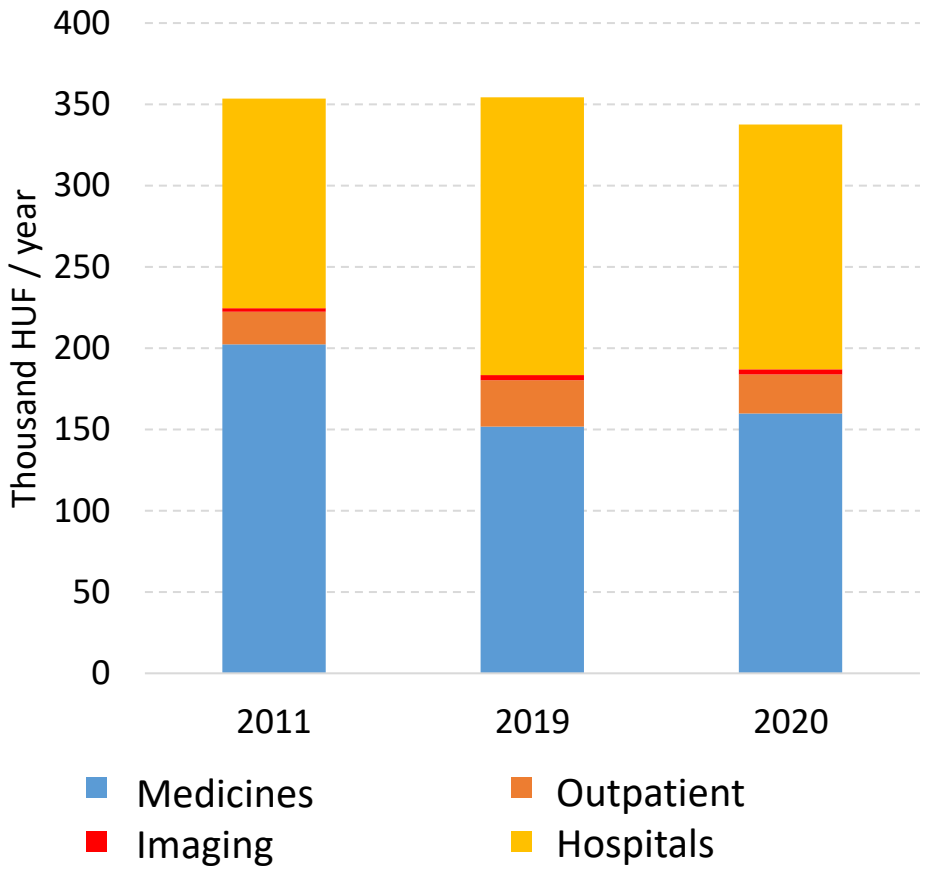
	Medicines	Outpatient services	CT/MRI imaging	Hospitals
2011	202	20	2,1	129
2019	178	33	3,8	200
2020	193	29	3,6	182



Discharge records

Inflation corrected health insurance fund payments per capita (thousand HUF a year)

	Medicines	Outpatient services	CT/MRI imaging	Hospitals
2011	202	20	2.1	129
2019	152	28	3.3	171
2020	160	24	3.0	151



Discharge records

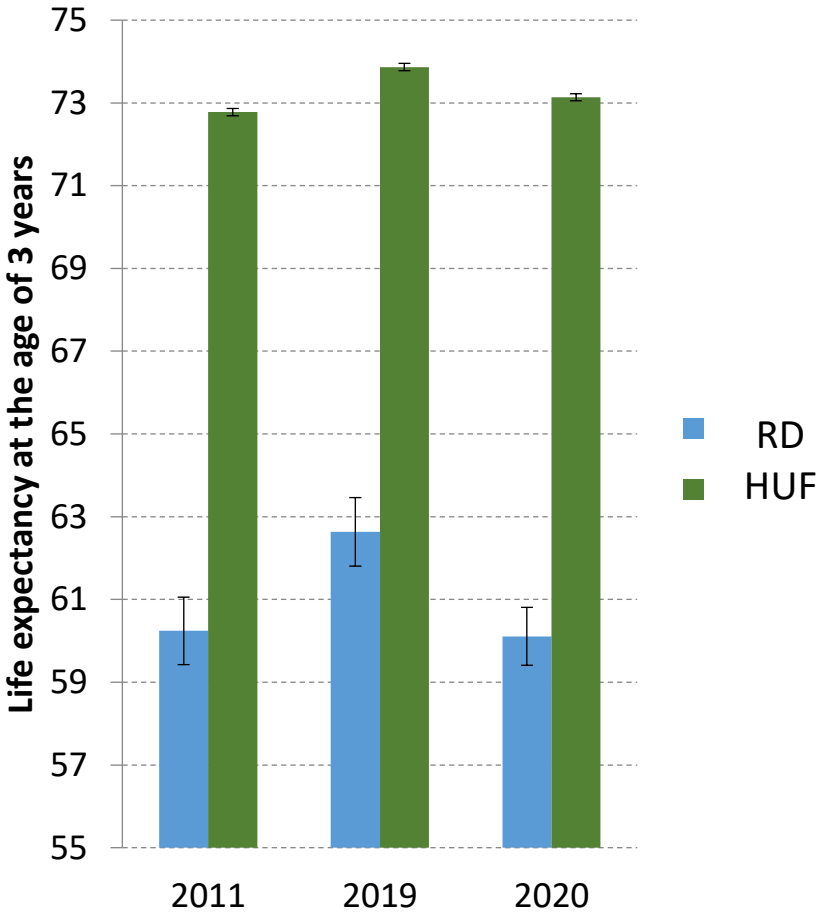
Lethal outcome

	Cases	Case fatality ratio	Change
2011	4668	3.02%	REF
2019	4956	2.88%	96%
2020	7275	3.63%	120%

Discharge records

Life expectancy at 3y

	2011	2019	2020
RD patients	60.2 y	62.6 y	60.1 y
Hungary	72.8 y	73.9 y	73.1 y
Difference	12.5 y	11.2 y	13.0 y
Change of difference	REF	-1.3 y	0.5 y



Proposed indicators for 1/3 of the RD patients by ICD10 codes

Cause of death data (Central Statistical Office)

- Relative importance of rare disease mortality
- Standardized death rate
- Proportion of death during the first year of life
- Potential years of life lost attributed to rare diseases
- Mean age at death
- Mean at death by distance from the center of expertise
- Mean age at death by calendar year
- Change of diagnostic ability index

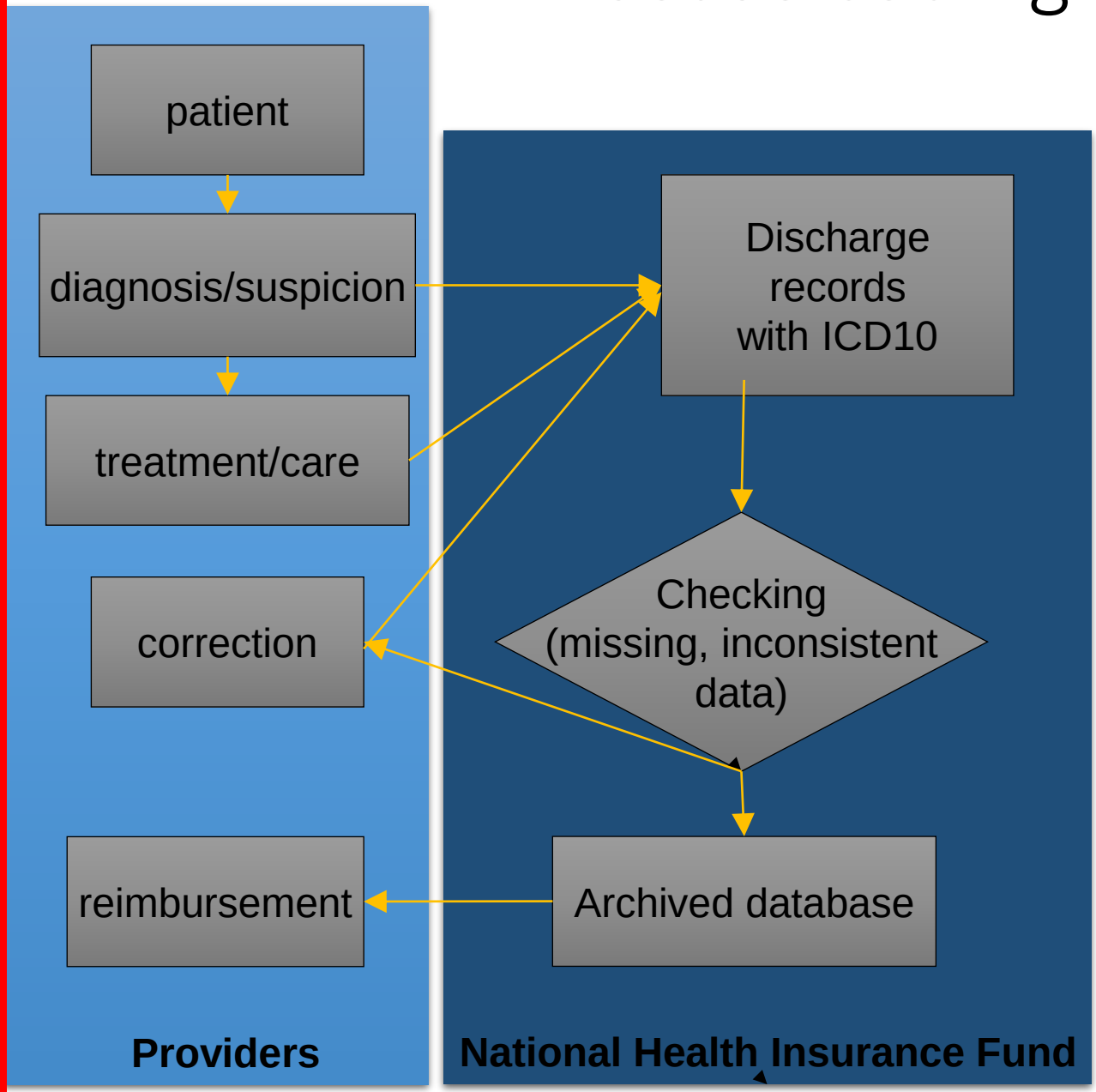
Discharge records of secondary care (National Health Insurance Fund)

- Prevalence of rare diseases
- Number of centers providing care for 50% of episodes
- Proportion of patients living in 50 km vicinity of the nearest center
- Visiting GP/outpatient/imaging/hospital a year
- Using outpatient/imaging/hospital services a year
- Payments for outpatient/imaging/hospital/medications a year
- Case fatality ratio, life expectancy

Automated cause of death coding

- The U.S. developed an automated system that captures this additional information and produces analyzable output based on it (began with 1968 data)
- Hungarian introduction: 2005

Disease coding in discharge records



Disease coding in discharge records + ORPHA coding