

Case Study in Huntington's Disease Burden

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Prof. Mondher Toumi MD, PhD.
Public Health Department
Aix Marseille University

Huntington's Disease Specificity

HD (Huntington's Disease) is a rare complex condition **3** main groups of symptoms. Each group is associated to different types of disturbance:

- **Motor disorder** (Chorea, voluntary movements, rigidity)
- **Cognitive disorder** (Spatial, memory)
- **Mental disorder** (Anxiety, Depression, Psychotics disorders)

No information on Burden

- Very few and very old studies on economic evaluation: partial and biased

- No information on QoL

- Patient pathology not described



No available data on **burden of illness** in any country.

No existing disease **framework model** on HD.

Burden of Illness Study

It is critical to document burden of illness objectively.

Objectives

- To obtain quickly data on cost of illness in EU-5 (UK, Italy, France, Spain, Germany) and Sweden → To have solid data for strategic messaging.
- To generate comprehensive high quality data
- To operate with limited budget.

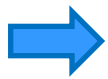
Gain Patient Groups Support

Launching of European Huntington's disease burden study (**Euro-HBD**), the first large comprehensive European study on the burden of HD.

- An international, observational, cross-sectional study.
- With the support of Patient group

Hurdles

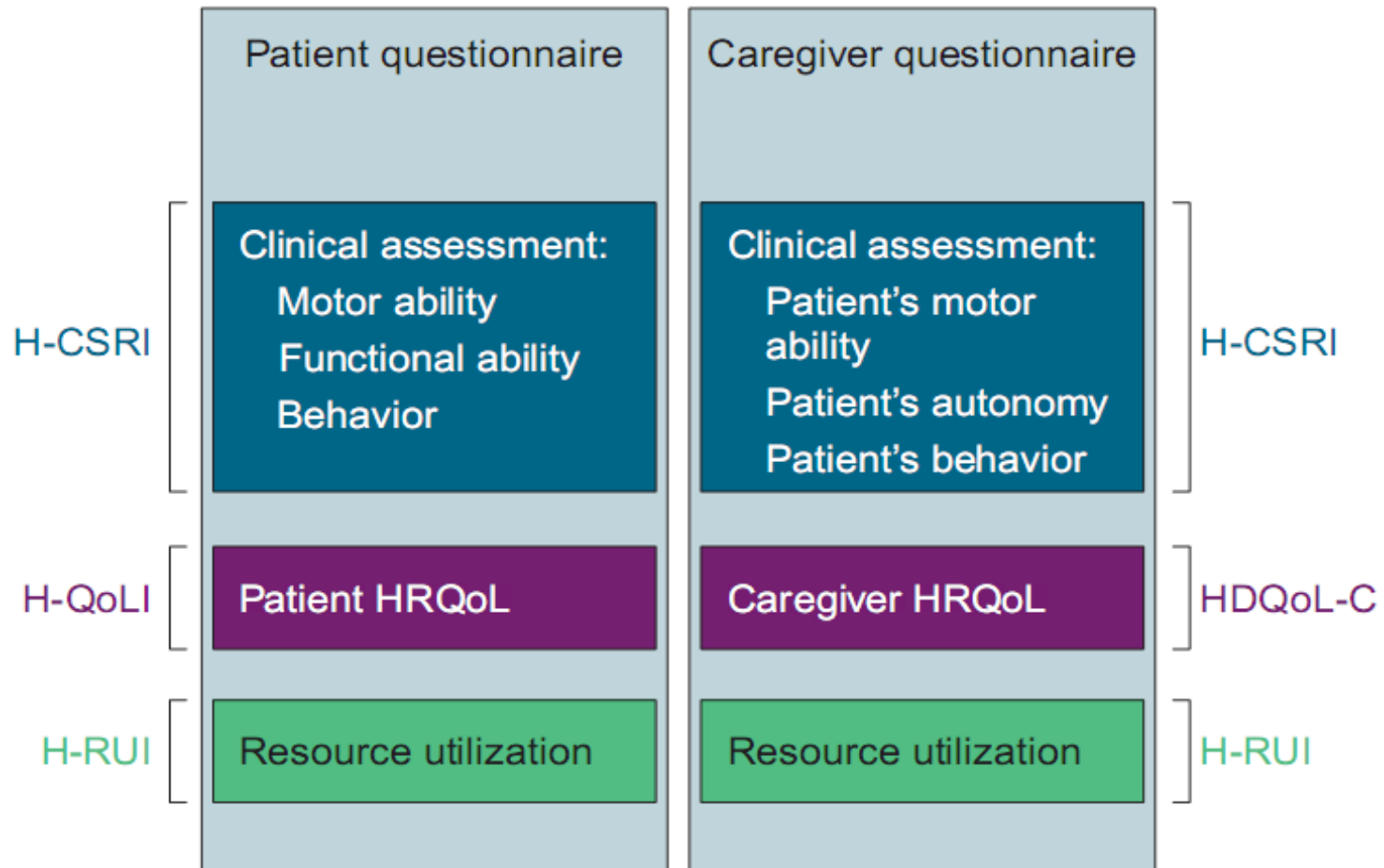
- New hurdles
 - Working with associations;
 - Gain trust;
 - Ensuring confidentiality of patients;
 - No available self-reported instrument;



Construction of a self-reported instrument

The Instrument

The Huntington Self-Assessment Instrument (HSAI)



Dysarthria: Physician reported

Patient questionnaire

Clinical Self-Reported Instrument (H-CSRI)

Motor disorder evaluation

- Example 1: **DYSARTHRIA**
 - 0 Normal
 - 1 Unclear, no need to repeat
 - 2 Must repeat to be understood
 - 3 Mostly incomprehensible
 - 4 Anarthria

Dysarthria: Patient reported

Construction of the instrument

1. How easy is it for you to say single words which are understood by others?

0	1	2	3	4
I have no problem in saying a single word and being understood	A little difficulty	Moderate difficulty	Severe difficulty	I cannot say a word, or people do not understand my words

2. How easy is it for you to say sentences (speech), which are understood by others?

0	1	2	3	4
I have no problem in speaking and being understood	A little difficulty	Moderate difficulty	Severe difficulty	My speech is severely impaired and most of time, people do not understand me

Adiadokokinesis: Physician reported

Patient questionnaire

Clinical Self-Reported Instrument (H-CSRI)

Motor disorder evaluation

■ Example 2: **PRONATE/SUPINATE - HANDS**

- | | |
|---|--------------------------------|
| 0 | Normal |
| 1 | Mild slowing and/or irregular |
| 2 | Moderate slowing and irregular |
| 3 | Severe slowing and irregular |
| 4 | Cannot perform |

right

left

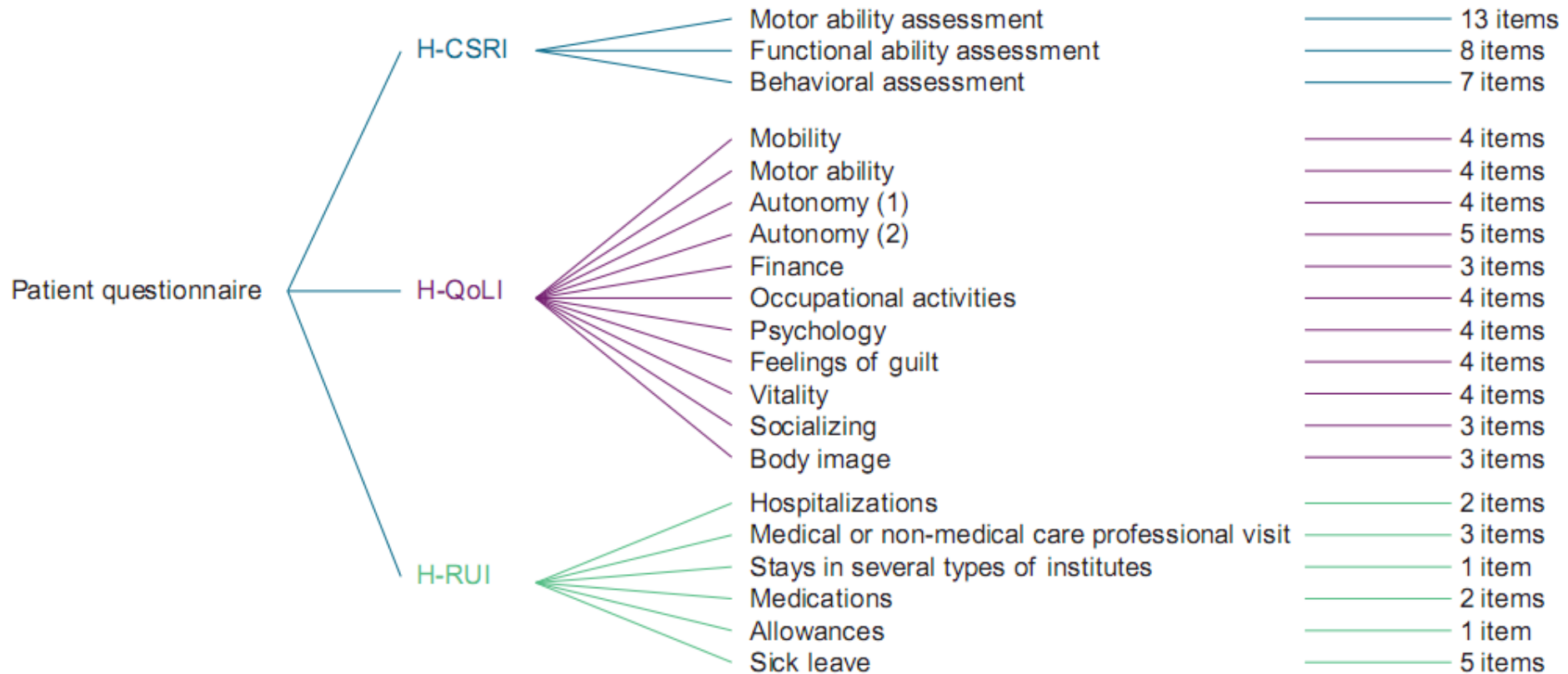
Adiadokokinesia : Patient reported

Construction of the instrument

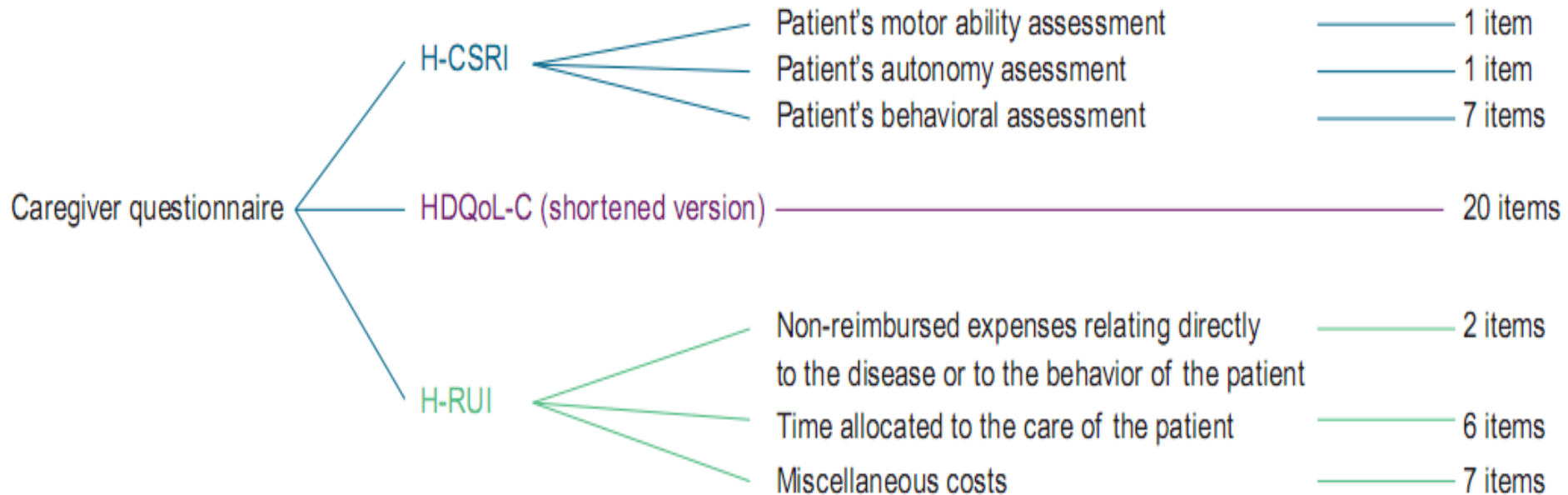
11. In your opinion, are you able to open a locked door by putting a key in a keyhole and turning it?

	0	1	2	3	4
	In my opinion, I am able to do this task normally and without difficulty	With a little difficulty	With moderate difficulty	With severe difficulty	In my opinion, I am not able to do this task

Burden of Illness Study - Patient



Burden of Illness Study - Caregiver



Validation

The entire validation process of HSAI was needed and realized in parallel of the data collection

Within the study

- Internal validation

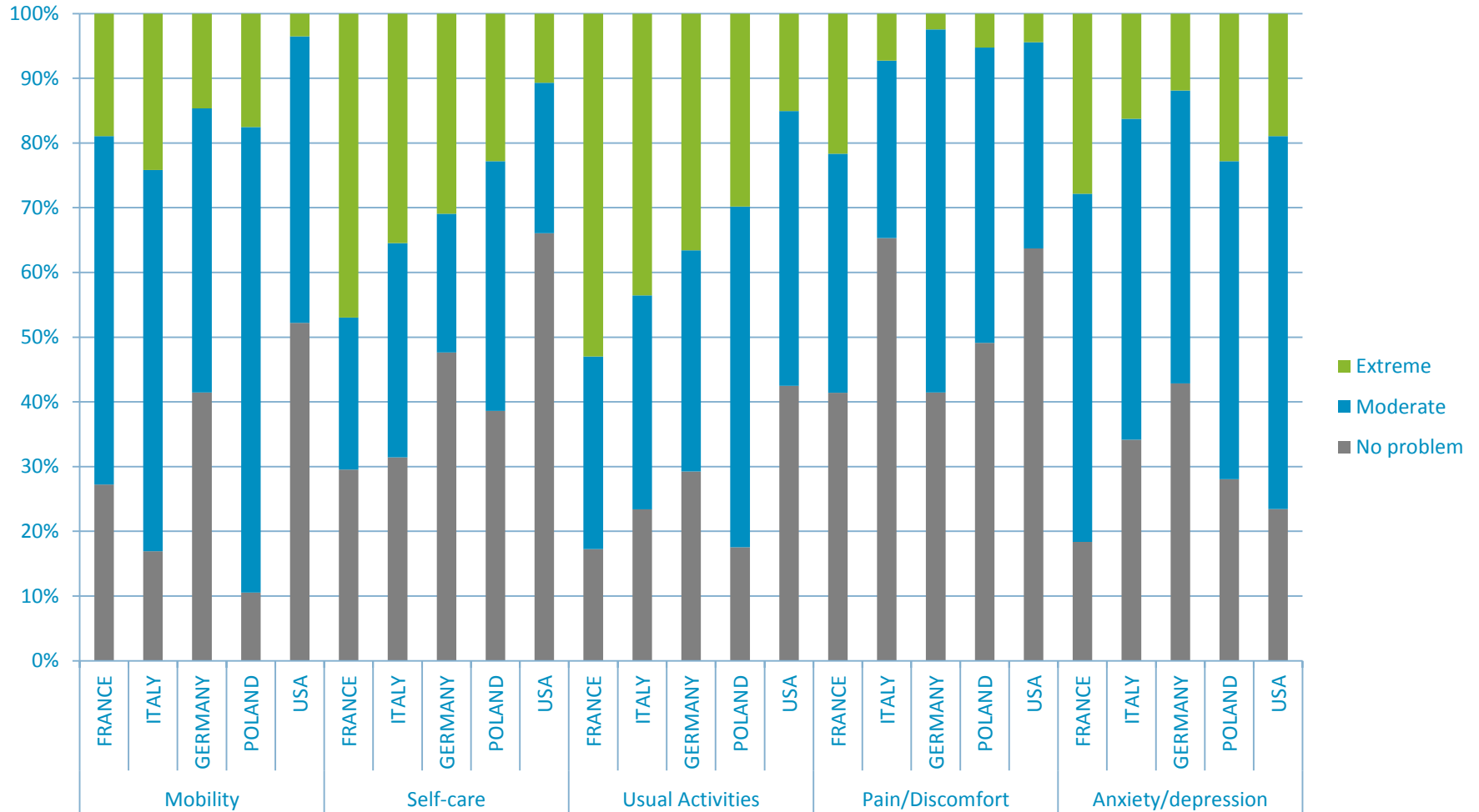
Outside the study

- External validation

Population Disposition

	France N = 176	Italy N = 124	Germany N = 44	Poland N = 60	The USA N = 165
Demographic					
Male, n (%)*	88 (50%)	58 (47%)	20 (45%)	25 (42%)	54 (33%)
Age, years*	57 (11)	54 (12)	50.95 (13.95)	48.02 (13.06)	49.07 (13.36)
Disease history					
Age at onset of symptoms, years*	44 (13)	45 (11)	40.55 (13.60)	38.02 (15.55)	40.53 (15.20)
Age at clinical diagnosis, years*	46 (13)	47 (11)	42.57 (14.22)	44.25 (12.91)	43.11 (14.88)
Tested genetically, n (%)	143 (81%)	114 (92%)	40 (93%)	52 (90%)	122 (83%)
Motor score					
Patient motor score*	36.77 (20.38)	35.95 (19.72)	31.46 (17.72)	31.34 (16.91)	20.65 (15.53)
Patient chorea score*	7.37 (4.86)	7.31 (4.92)	7.81 (4.59)	6.75 (3.99)	4.83 (4.10)
Modified motor score*	30.05 (16.91)	28.65 (15.74)	24.33 (14.78)	24.24 (13.75)	16.71 (13.33)
Behavioural score					
Depression-anxiety*	11.60 (5.169)	10.36 (6.04)	8.74 (5.01)	9.31(6.04)	10.52 (5.19)
Impulsivity	4.05 (2.39)	3.58 (2.73)	3.03 (2.10)	3.98 (2.50)	3.63 (2.32)
Psychotic disorder*	2.62 (2.81)	2.21 (2.80)	NA	1.46 (1.80)	NA
Cognition*	5.74 (3.89)	4.39 (4.06)	NA	3.98 (3.94)	NA
Functional score*	4.92 (2.36)	4.47 (2.53)	4.21 (2.40)	3.51 (1.96)	2.65 (2.5)

EQ5D Results



Results

	France (N =176)	Italy (N = 124)
Consultation	750 (1140)	540 (543)
Paramedical	3362 (4242)	567 (1238)
Services	4042 (8177)	1120 (3261)
Hospitalization	8368 (11632)	1680 (4412)
Facility	10 579 (22759)	1925 (11387)
Sick leave	2740 (13348)	523 (4596)
Total patient cost	30 572 (34 212)	6461 (14 524)
Caregiver burden (time involvement)	9.08 (15.03)	22.49 (24.94)
Caregiver burden (time involvement in cost)	80 (132)	152 (168)
Total caregiver cost	501 (1119)	408 (588)

Conclusion

Patient groups may represent an invaluable asset when conducting observational studies.

Conducting research with patient groups may be challenging

- Technically to **develop validated PRO instruments;**
- But also, for **accessing to patients and gain trust with associations.**

• **THANK YOU**