

1 Article

2 Exploring the reliability and validity of the 3 Huntington's Disease Quality of Life Battery for 4 Carers (HDQoL-C) within a Polish population

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20 **Abstract:** Huntington's disease (HD) is a rare genetic neurodegenerative disorder that causes
21 motor disorders, neuropsychiatric symptoms and a progressing deterioration of cognitive
22 functions. Complex issues resulting from the hereditary nature of HD, the complexity of symptoms
23 and the concealed onset of the disease have a great impact on the quality of life of family carers. The
24 caregivers are called as "forgotten people" in HD family, also in genetic counseling. This study
25 aims to explore the reliability and validity of the Huntington's Disease Quality of Life Battery for
26 carers (HDQoL-C) within a Polish population. 90 carers recruited from the Enroll-HD study in
27 Polish research centres of the European Huntington's Disease Network completed a Polish
28 translation of the HDQoL-C. Data was subjected to Principle Components Analysis and reliability
29 measures. The Polish version of the shortened versions of the HDQoL-C is similarly valid compared
30 to the original English version and suitable for use within this population. The HDQoL-C has
31 previously demonstrated a wide range of benefits for practitioners in capturing and
32 understanding carer experience and these benefits can now be extended to Polish speaking
33 populations.

34 **Keywords:** Huntington's disease; quality of life; family caregiving; reliability and validity; factor
35 analysis; Poland

36

37 1. Introduction

38 Huntington's disease (HD) is a rare genetic neurodegenerative disorder caused by the mutation
39 of the IT15 gene which codes the huntingtin protein located on the short arm of chromosome 4 [1].
40 The clinical presentation of the disease includes motor disorders, neuropsychiatric symptoms and a
41 progressing deterioration of cognitive functions. The motor symptoms consist of involuntary
42 choreatic movements and impaired saccadic eye movements. The symptoms that appear at the
43 subsequent stages of the disease are dystonia, dysarthria, dysphagia, rigidity and bradykinesia,
44 leading to death in 15-20 years [2,3]. The most frequent causes of death are aspiration pneumonia,

45 injuries resulting from falls and suicides, which are recorded twice as often as in the total population
46 [4]. In the juvenile form, usually presenting itself with mental degradation, motor disorders occur
47 later, the course of the disease is more acute, followed by death after 8-10 years from the onset of the
48 first symptoms. The range of neuropsychiatric symptoms in HD is within 33-76%, of which the most
49 frequent are: depression, anxiety disorders, irritability, apathy, obsessive-compulsive disorders and
50 psychotic symptoms [5,6]. Cognitive disorders include memory deterioration, slowed down
51 thinking processes, disorders of executive and visual and spatial functions, problems with
52 organisation, planning and multitasking, difficulties with decision-making and with dealing with
53 new situations [7].

54 Despite the fact that diagnosing HD is traditionally based on motor symptoms, the highest
55 impact on patients and carers is exerted by mental state and behaviour changes [8,9] as well as
56 cognitive disorders, which may appear as early as 12-15 years before the diagnosis and delay the
57 accurate diagnosis of HD [10,11].

58 Due to the autosomal dominant type of inheritance HD is passed over from generation to generation
59 and has damaging effects both on the patients and their carers/families, who usually become
60 responsible for caring for the family member with HD [12]. The disease's trajectory in the family may
61 last up to 30 years or even more [13]. The disease being passed from generation to generation leads
62 to situations when for each patient there are 20 people affected by the disease in various ways, such
63 as the risk of HD, issues related to care or the effects of the disease on the family [14].

64 Complex issues resulting from the hereditary nature of HD, the complexity of symptoms and the
65 concealed onset of the disease [15] have a great impact on the quality of life of family carers [16],
66 who are aware that the disease might develop in other family members. Carers blame themselves for
67 passing the disease on to the next generation [13,17-19].

68 For each child in a family with a parent having HD the risk of inheriting the gene is 50%, [12] while
69 diagnostic and preclinical trials clearly confirm the genetic status of a person [1]. Carers fear that
70 they might be forced to provide care for several generations, for a few people at the same time, and
71 such a situation might last for years [20].

72 There are few studies which directly investigate the impact of HD on the quality of life (QoL) of
73 family carers [21]. The existing studies demonstrate that the quality of life of carers is seriously
74 compromised due to their burden [22] these capture a unique sense of long-term isolation and
75 frustration [23] the performed role and conflicting roles, on the one hand requiring caring for the
76 partner and, on the other, for the children, who they need to protect from unexpected behaviour,
77 irritability or aggression of the HD patient [24], insufficient resources to perform the care duties is
78 also of essence [25-27]. Carers experience substantial changes in communication with HD patients
79 [28] and their ability to perform care and parental functions deteriorates as the disease progresses
80 [29,30] and as their physical and mental health declines [18].

81 The quality of life of carers is considerably reduced due to the lack of access to specialised
82 medical care, while medical personnel are often unaware of the immense impact of HD on the family
83 and unprepared to solve specific problems resulting from the complex changes in HD families [31].
84 Feeling abandoned by the system, carers describe their experiences with medical professionals as "a
85 lone journey" [27,32]. Family carers were described as "the forgotten" both in the families with HD
86 [33], and in the genetic counselling system [34].

87 HD is currently incurable and there are no effective treatment methods, the concept of the quality of
88 life is particularly important due to the unique burden carried by carers. Using specific
89 questionnaires to evaluate the QoL of carers makes it possible for physicians and researchers to
90 assess the emotional and physical functioning and lifestyle from the carer's perspective, and does
91 not provide an objective interpretation [21]. It offers a possibility to assess changes in the QoL of
92 carers over time, especially as despite many promising treatment-options, the disease is currently
93 incurable, and one thing is certain: the path towards the solution is long [35].

94 The existing literature highlights the burden that family carers may face in supporting a loved
95 one living with HD but as yet there is no validated quality of life measure to assess the impact of

96 caring on the QoL of family members caring for a loved one with HD in Poland. In Poland there is a
97 lack of studies on the impact of HD on the quality of life of family carers.

98 According to our knowledge, this is the first study with the participation of Polish family carers
99 on such a numerous group with the use of a specific scale for evaluating the quality of life of carers
100 in HD. Thus far only two reports referring to Polish carers of HD patients have been produced
101 [26,36].

102 The aim of this study was therefore to validate a translated version of the Huntington's Disease
103 Quality of Life battery for carers (HDQoL-C)[16] for use in Poland.

104 2. Materials and Methods

105 2.1. Instrumentation

106 The HDQoL-C is a multidimensional, disease-specific and subjective health related quality of
107 life tool that incorporates the individual's physical health, psychological state, level of
108 independence, social relationships and personal beliefs. The scale was developed as an outcome
109 measure and in addition can be used to assess subjective QoL in family carers of people with HD.
110 Cronbach's alpha scores for the three components of the original HDQoL-C scale demonstrate good
111 internal consistency - 0.801 (Practical aspects of caregiving); 0.844 (Satisfaction with life) and 0.885
112 (Feelings about living with HD), with test re-test reliability for the same components being 0.86; 0.90;
113 0.92 respectively [16]. The scale demonstrates good congruent validity, good face validity and robust
114 content validity. It has been translated and validated with success into French and Italian also[21].
115 The HDQoL-C is being used in ongoing Euro-Huntington's Disease Burden studies measuring the
116 impact of Huntington's Disease in several European countries and in the USA. To develop the Polish
117 version of the HDQoL-C, forward-backwards translation methods were applied to the original
118 English version of the scale. The translation did not show any errors in translation.

119 2.2. Participant and Procedure

120 Participants were recruited among carers taking part in the Enroll-HD study in Polish research
121 centres of the European Huntington's Disease Network. Contact with carers was also established via
122 the Polish Huntington's Disease Association during the annual conference in Warsaw, Poland
123 devoted to HD, via carers involved in adding new members to the surveyed group, and through the
124 "forum - HD zamki" website. The selection criteria of carers for the study was age ≥ 18 years, the
125 presence of a patient living at home and written consent to participate in the study. All carers who
126 took part in the study had a loved one under their care who had tested positive for HD.
127 From 100 carers contacted directly, through post or electronic mail, 90 responded to the invitation to
128 take part in the study, which was carried out from June 2015 to December 2016. 18 of 20 carers
129 answered a re-test questionnaire after a 2-week interval to gather data on test-re-test reliability.
130 Because, HD is a rare disease and there is no detailed data indicating the prevalence of disease in
131 Poland, the sample size is likely to be representative for the aim of this study. Ethical approval to
132 conduct this study was granted by the Bioethics Committee of the Medical University of Lublin,
133 Poland, (Protocol number KE-0254/134/2015). A written informed consent was obtained from each
134 participant.

135 2.3. Data Analysis

136 Principal components analysis (PCA) was used to explore the inter-relationship between the
137 variables on the HDQoL-C. Bartlett's test of sphericity and the Kaiser-Meyer-Olkin (KMO) measure
138 of sampling adequacy were also used to assess the suitability of the data for PCA.
139 The Cronbach's α coefficient was used to assess the internal consistency of the scale. A reliability
140 threshold level was considered acceptable when greater than 0,70. Reproducibility assesses if an
141 instrument produces the same results on repeated administrations when respondents have not

142 changed. The reliability coefficient was computed by correlating instrument scores for the two
143 administrations.

144 3. Results

145 3.1. Respondent Characteristics

146
147 The sample comprised 90 carers. 57.8 % of carers were the main carers of a HD patient. The
148 carer ages ranged between 20 and 80 years, with a mean of 48.78 years. Most carers were women
149 (68%) and weren't carers before (81%). 57.8 % carers were married and 41.1% had a child with a risk
150 of disease. Mean length of caring for an HD affected family member was 7.81 years. Table 1 presents
151 the characteristics carers in family with HD.

152

153 **Table 1.** Characteristic of the researched group family carers (n= 90).

Characteristics of family carers	N	%
Women	61	68
Age (years)		48.78 (\pm 15.21)
Main carer	52	57.8
Marital status		
Married	52	57.8
Single	17	18.9
Widowed	13	14.4
Partnership	5	5.6
Divorced	3	3.3
Family situation		
Number of years since HD knowledge in family		11.76 (\pm 10.4)
Have children at risk	37	41.1
Relation with HD patient		
Husband/wife	31	34.4
Parent	26	28.9
Child	14	15.6
Other	10	11
Sibling	8	8.9
Partner	1	1.2
Carer background		
Carer has previously cared any other HD affected person	17	19
Duration of caring (in years)		7.81 (\pm 8.48)

154

155

156 3.2. Principle Components Analysis and Reliability

157

158 In line with the English language version, separate Principle Components Analyses were
159 conducted. Section One of the questionnaire is comprised of demographic information and so was
160 not included in the analysis. For Section Two the Kaiser-Meyer Olkin measure of sampling adequacy
161 showed that the sample was factorable (KMO=. 714). Bartlett's Test of Sphericity was highly
162 significant ($\chi^2 = 210.4$, $df=36$, $p<.001$), and low off-diagonal values in the anti-image correlation
163 matrix demonstrated that the data were suitable for factor analysis [37].The analysis revealed two
164 factors. The cross loadings displayed for item QoL2_2 in Factor 1 may indicate cause for concern as it

165 also negatively loaded on to factor 2 at $-.334$. Table 2 outlines the Pattern Matrix of rotated factor
 166 loadings for section 2.

167

168 **Table 2.** Pattern Matrix of rotated factor loadings for section 2.

169

Item	Content	Factor	
		1	2
QoL2_3	How often do you have access to professionals that have specialized knowledge of HD and understand its implications?	.827	-.072
QoL2_4	How much support are you given by health care professionals?	.788	-.030
QoL2_6	How often do you have access to appropriate care facilities?	.766	.004
QoL2_7	How often do you receive any practical support you need?	.754	.036
QoL2_2	How often do you receive appropriate help from social services?	.610	-.334
QoL2_1	How often are you restricted by the need to maintain a regimented daily routine?	.039	.816
QoL2_8	How often do you experience a conflict of interest between what you want and what your HD affected relative wants?	.067	.741
QoL2_5	How often does the inherited nature of HD further complicate your caring role?	-.117	.667
QoL2_9	How often do you sleep well?	-.087	.337

170

171 Internal consistency was analysed using Cronbach's Alpha. The items in Factor 1 demonstrated high
 172 reliability (Cronbach's $\alpha = .81$). Factor 2 demonstrated moderate reliability (Cronbach's $\alpha = .58$), which
 173 increased to $\alpha = .64$ if item QoL2_9 was deleted.

174 The PCA for Section 3 showed a Kaiser-Meyer Olkin measure of sampling adequacy indicating that
 175 the sample was factorable (KMO = .858). Bartlett's Test of Sphericity was highly significant ($\chi^2 =$
 176 354.3 , $df=28$, $p<.001$), and low off-diagonal values in the anti-image correlation matrix demonstrated
 177 that the data were suitable for factor analysis. The analysis produced a two-factor solution, however
 178 the second factor only featured a single item (QoL3_7). Table 3 outlines the Pattern Matrix of rotated
 179 factor loadings for section 3.

180

Table 3. Pattern Matrix of rotated factor loadings for section 3.

Item	Content	Factor	
		1	2
QoL3_2	How satisfied are you with what you achieve in life?	.860	-.128
QoL3_8	How satisfied are you with your overall quality of life?	.825	.246
QoL3_4	How satisfied are you with how safe you feel?	.811	.223
QoL3_1	How satisfied are you with your health?	.792	.000

Qo13_6	How satisfied are you with your own happiness?	.763	.329
Qo13_5	How satisfied are you with feeling a part of your community?	.656	.373
Qo13_3	How satisfied are you with your close relationships with family or friends?	.566	.464
Qo13_7	How satisfied are you with the treatment that your HD affected relative receives?	.019	.926

181

182 The items in Factor 1 demonstrated high reliability (Cronbach's $\alpha = .90$).

183 PCA for section 4 indicated a Kaiser-Meyer Olkin measure of sampling adequacy such that the
 184 sample was factorable (KMO=.795). Bartlett's Test of Sphericity was highly significant ($\chi^2 = 569.2$,
 185 $df=136$, $p<.001$), and low off-diagonal values in the anti-image correlation matrix demonstrated that
 186 the data were suitable for factor analysis [38]. A four-factor solution was indicated, however, the
 187 items in the fourth factor were problematic with high cross loadings. Table 4 outlines the Pattern
 188 Matrix of rotated factor loadings for section 4.

189

190

Table 4. Pattern Matrix of rotated factor loadings for section 4

191

Item	Content	Factor			
		1	2	3	4
Qo14_7	I feel sad or depressed	.775	.052	.310	.097
Qo14_17	I feel like I don't know who I am anymore	.765	.087	-.017	.006
Qo14_8	I feel stressed	.749	.051	.143	.129
Qo14_5	I feel exhausted	.729	.126	.186	-.161
Qo14_16	I feel that i have had a "duty of care" forced on me	.711	-.166	-.269	.213
Qo14_10	I feel my own needs are not important to others	.645	-.063	.400	-.157
Qo14_3	I feel isolated	.592	.213	.282	-.154
Qo14_4	I feel there is hope for the future	.036	.797	.287	.004
Qo14_11	I feel comforted by the belief that one day there will be a cure for HD	.024	.773	-.155	.160

QoL4_13	I feel comforted by my beliefs	.060	.674	.084	.253
QoL4_9	I feel worried about the genetic consequences of HD	.038	-.024	.793	.131
QoL4_1	I feel guilty	.220	.080	.590	-.006
QoL4_6	I feel supported	.176	.386	.474	.125
QoL4_12	I feel that HD brought something positive to my life	-.029	.334	.076	.701
QoL4_15	I feel that HD has made me a stronger person	.166	.404	.246	.673
QoL4_2	I feel financially disadvantaged	.424	.179	.170	-.583
QoL4_14	I feel that I can cope	.282	.322	.467	.546

192

193 Internal consistency was again analysed using Cronbach's Alpha. The items in Factor 1
 194 demonstrated high reliability (Cronbach's $\alpha = .85$). Factor 2 demonstrated -good reliability
 195 (Cronbach's $\alpha = .70$), Factor 3 had moderate reliability (Cronbach's $\alpha = .51$). Factor 4 was moderately
 196 reliable (Cronbach's $\alpha = .56$), however, if item QoL4_2 was excluded the reliability of this sub-scale
 197 increased considerably (Cronbach's $\alpha = .78$).

198

199 *3.3. Test-Re-test*

200

201 18 caregivers filled questionnaire again after 2 weeks. Coefficients are presented in the Table 5. All
 202 components present high statistically significant correlation ($p < 0.001$) with satisfying coefficient of
 203 determination ($r^2 > 0,6$).

204

Table 5. Test-retest correlations for Sub-scales

Sub-scales	II	III	IV
II	0.82	-	-
III	-	0.92	-
IV	-	-	0.83

205

206 **4. Discussion**

207 By measuring QoL in this Polish population we were able to build on our understanding of the
 208 issues surrounding caregiving in HD in order to establish ways of improving QoL for this carer
 209 group. The translation of the HDQoL-C into an additional language means the scale can be used
 210 even more widely, allowing for further comparisons across Europe. The need to translate and adapt
 211 QoL instruments for use in languages other than the source language (usually English) has increased

212 with the internationalization of clinical trial programmes and cross-cultural research. For example,
213 the ISPOR Task Force[37]note the importance of evidencing similarities in measurement properties
214 between all versions of the same tool to pool analysis and facilitate comparability between countries.
215 The HDQoL-C, has previously been translated into French and Italian [21] and the addition of this
216 Polish translation has the potential to further our understanding of any cross-cultural differences in
217 the resources, policies and practices that may influence the QoL HD family carers on a global level.
218 The Polish version of the shortened versions of the HDQoL-C is similarly valid compared to the
219 original English version. The scale instrument showed satisfactory face validity with little missing
220 data (1,1%). The Cronbach's alpha coefficients demonstrate moderate to good reliability. There were
221 two items, "How often do you sleep well?" (section 2, question 9) and "I feel financially
222 disadvantaged" (section 4, question), that if removed increased the reliability of the subsection they
223 sit within. The question "How often do you sleep well?", has consistently reduced factor reliability
224 even in the original version and was kept in due to the emphasis that carers have placed on it despite
225 not rotating well in the factor analysis. It may be that this question needs refinement in terms of
226 wording to improve reliability or that the subjectivity of this item is difficult to articulate. With
227 regards to the statement "I feel financially disadvantaged" it may be that this statement does not
228 translate cross culturally or is not relevant for a Polish population. It may also be the case that the
229 Polish carers were less focused on financial disadvantage than on the heritable or familiar elements
230 of their experience of caring for someone with HD.

231 It should also be emphasised that the contact with carers during collecting data for the study was of
232 a unique nature and had a therapeutic effect on them. They expressed that they were pleased that
233 their roles were recognised, as most measures focus on HD patients. This is in line with evidence
234 from [16] who also observed a cathartic benefit of engaging with family carers, many of whom
235 described completing the questionnaire as an intervention itself.

236 A lack of items bias in English and Polish translations confirms the scale's multi-lingual,
237 multi-cultural consistency and indicates that the scale is easily applicable in other languages. The
238 Polish version of the HDQoL-C demonstrated good internal consistency and congruent validity.
239 Further validation, such as test-retest validity and sensitivity to changes, would enhance this
240 validation process.
241

242 5. Study limitations

243 Although this is the first study in Poland with the use of the HD-specific quality of life
244 questionnaire for family carers, it has some limitations. The group of carers was quite small (90
245 family carers, only 10 did not respond to the invitation to study) due to the rare occurrence of HD. It
246 should be emphasized that access to individual caregivers is difficult and it is only possible during a
247 conference that is organized annually by the Polish Huntington's Disease Association (only those
248 who benefit from such support) and an online forum during which there is no possibility of direct
249 conversation. The most important thing, however, is that during conversations at the conference and
250 in subsequent telephone conversations, the carers emphasized that just filling out the questionnaire
251 and contacting researchers was a positive experience for them, thanks to which they had the
252 opportunity to verbalize their feelings and difficulties related to the role of a caregiver. This is
253 confirmed by the comments at the end of the questionnaire, in which the caregivers thanked for
254 noticing them and understanding. At the same time, they emphasized that they do not have the
255 closest people with whom they could talk, because the family avoids topics about HD. Of the entire
256 study group, only 7 people did not leave contact, which also indicates their needs. The group of 90
257 respondents was also created thanks to carers who willingly gave contact to other caregivers.
258

259

260

261 6. Practice Implications

262 Interest in the QoL of carers is crucial due to the practical, socio-economic aspects. In the
 263 situation when care-related costs are borne mainly by carers and not the State budget, incapacities of
 264 carers generate unplanned expenses for medical and social assistance [39]. Due to the fact that carers
 265 feel like a forgotten group in HD families, it is necessary to promote quality of life in primary
 266 healthcare. The time of caring for a patient with HD is much longer than in other neurodegenerative
 267 diseases and the need for environmental care is also longer. It is recommended to study the quality
 268 of life of HD caregivers in primary healthcare in order to implement appropriate support
 269 procedures at various stages of the disease, the more so that carers can also get ill and pass defective
 270 genes to their children.

271 7. Research Recommendations

272 This study is part of a wider project on family carers in HD in Poland. Further research, with
 273 quantitative and qualitative approach, can identify other areas of quality of life for HD families.
 274 Moreover, due to the multitude of symptoms (multifaceted disease) conducting interdisciplinary
 275 research would allow a better understanding of the needs of family carers and this would translate
 276 into providing them with practical support. Therefore, further research based on mixed
 277 methodology and done within multidisciplinary teams is recommended.
 278

279 8. Conclusions

280 A lack of item bias in English and Polish translations confirms the scale's multi-lingual,
 281 multi-cultural consistency and indicates that the scale is easily applicable in other languages. The
 282 Polish version of the HDQoL-C demonstrated good internal consistency and congruent validity.
 283 Further validation, such as sensitivity to changes, would enhance this validation process. The
 284 HDQoL-C has demonstrated a wide range of benefits for practitioners in capturing and
 285 understanding carer experience and these benefits can now confidently be extended to Polish
 286 speaking populations.

287 **Availability of data and materials:** The HDQoL-C is the property of Dr Aimee Aubeeluck, CPsychol, FHEA.
 288 It has been developed for use by family members, researchers and clinicians and can be used and adapted
 289 freely for the benefit of improving the quality of life of families living with Huntington's Disease. To use the
 290 scale, contact is required: aimee.aubeeluck@nottingham.ac.uk and cite in any subsequent write up.

291 **Author Contributions:** The co-authors had together contributed to the completion of this article. Specifically, it
 292 follows their individual contribution: Conceptualization, A.B. and A.A.; Methodology, E.S.; Ad.B.; Data
 293 curation, A.B., Ad.B.; Project administration, Ad.B. and K.K.; Supervision, A.A. and B.S.; Writing—original
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