Supplementary Table 1. C-GUIDE scores by 17 utility items (n=247)

C-GUIDE items for primary indication for testing*^	Number of responses (%) (n=247)	Mean C-GUIDE score (SD) (n=245)[+]
1. Provided a genetic explanation for my patient's health condition		
Provided a COMPLETE genetic explanation [2]	59 (24%)	0.55 (0.05)
Provided a PARTIAL genetic explanation [1]	5 (2%)	
Provided a POSSIBLE genetic explanation [1]	13 (5%)	
Provided NO genetic explanation [0]	170 (69%)	
2. Reduced the likelihood of other potential diagnoses in my differential		
COMPLETELY REDUCED the likelihood of other potential diagnoses in my differential [2]	59 (24%)	0.51 (0.05)
PARTIALLY REDUCED the likelihood of other potential diagnoses in my differential [1]	9 (4%)	
DID NOT REDUCE the likelihood of other potential diagnoses in my differential [0]	168 (68%)	
Not appliable [0]	11 (4%)	
3. Provided information about natural history		
Provided SIGNIFICANT information about the natural history of or medical issues associated with my patient's condition [2]	60 (24%)	0.54 (0.05)
Provided SOME information about the natural history of or medical issues associated with my patient's condition [1]	14 (6%)	
Provided NO information about the natural history of or medical issues associated with my patient's condition [0]	173 (70%)	
4. Indicated further testing to identify a genetic diagnosis can be avoided		
Indicated that further testing to identify a genetic diagnosis CAN BE AVOIDED [2]	61 (25%)	0.49 (0.06)
Indicated that further testing to identify a genetic diagnosis MAY STILL BE REQUIRED, now or in the future [0]	186 (75%)	
5. Indicated that previous surveillance or monitoring related to my patient's condition can be discontinued or avoided^		
Indicated that previous surveillance/monitoring can be DISCONTINUED OR AVOIDED [2]	0 (0%)	0.0 (N/A)
Indicated that previous surveillance/monitoring is STILL REQUIRED [0]	239 (97%)	
Previous surveillance/monitoring is NOT RELEVANT to this case [0]	6 (2%)	
6. Facilitated my patient's access to or continuation of a community or educational service (e.g. learning, rehabilitation resources) that would not have been available without the testing^		
FACILITATED access to or continuation of a community or educational service [2]	64 (26%)	0.52 (0.06)
DID NOT FACILITATE access to or continuation of a community or educational service [0]	181 (73%)	

7. Enabled me to identify and access a research study that I wouldn't have been able to access without the testing^		
ENABLED me to IDENTIFY and ACCESS a clinical trial [2]	34 (14%)	
ENABLED me to IDENTIFY a clinical trial [1]	26 (11%)	
Enabled me to IDENTIFY and/or ACCESS a natural history or functional study to assist with result interpretation [1]	1 (<1%)	0.39 (0.05)
DID NOT ENABLE me to IDENTIFY or ACCESS a clinical trial, natural history or functional study [0]	184 (74%)	
8. Enabled me to identify a support group for my patient or his/her family that I wouldn't have considered without the testing		
ENABLED me to identify a support group [2]	63 (26%)	0.51 (0.06)
DID NOT ENABLE me to identify a support group [0]	184 (74%)	
9. Prompted a referral or investigation for the purpose of surveillance or monitoring that would not have been prompted on clinical grounds^		
PROMPTED a referral or investigation for surveillance/monitoring [2]	33 (13%)	
PROMPTED a referral or investigation for surveillance/monitoring that MAY NOT BE NECESSARY (e.g. variant of uncertain significance) [1]	8 (3%)	0.30 (0.04)
DID NOT PROMPT a referral/investigation for surveillance/monitoring [0]	204 (83%)	
10. Provided information to guide medication management^		
GUIDED current medication management [2]	48 (19%)	
MAY GUIDE medication management in the future [1]	16 (6%)	0.46 (0.05)
DID NOT PROVIDE information that would guide medication management, now or in the future [0]	181 (73%)	
11. Provided information about surgical management^		
ENABLED a discussion or offer of a surgical option [2]	5 (2%)	
AVOIDED a discussion or offer of a surgical option [1]	2 (1%)	0.05 (0.19)
A surgical option is NOT RELEVANT at this time or NOT RELATED to the genetic test results [0]	238 (96%)	
12. Provided information about a contraindicated behaviour (e.g. competitive sports)^		
ENABLED me to provide information about a contraindicated behaviour [2]	19 (8%)	0.16 (0.03)
Information about a contraindicated behaviour is NOT RELEVANT at this time [0]	226 (92%)	
13. Provided recurrence risk information for my patient^		
Provided recurrence risk information that is RELEVANT to my patient at this time [2]	48 (19%)	
Provided recurrence risk information that MAY BE RELEVANT to my patient in the future [1]	19 (8%)	0.47 (0.05)
Cannot be determined (e.g. variant of uncertain significance, did not provide information) [0]	178 (72%)	

14. Provided recurrence risk information for my patient's family		
Provided recurrence risk information that is RELEVANT to my patient's family at this time [2]	62 (25%)	0.52 (0.06)
Provided recurrence risk information that MAY BE RELEVANT to my patient's family in the future [1]	5 (2%)	
Cannot be determined (e.g. variant of uncertain significance, did not provide information) [0]	180 (73%)	
15. Clarified potential health risks for my patient's family		
CLARIFIED potential health risks for my patient's family [2]	64 (26%)	0.51 (0.06)
DID NOT CLARIFY health risks for my patient's family [1]	27 (11%)	
Cannot be determined (e.g. variant of uncertain significance, family member(s) did not receive testing or unknown if tested) [0]	156 (63%)	
16. Generated psychosocial benefit for my patient or his/her family		
SIGNIFICANT psychosocial benefit was experienced [2]	51 (21%)	0.98 (0.04)
MODERATE psychosocial benefit was experienced [1]	140 (57%)	
NO psychosocial benefit was experienced [0]	51 (21%)	
Cannot be determined [0]	5 (2%)	
17. Generated psychosocial concern for my patient or his/her family		
SIGNIFICANT psychosocial concern was experienced [-2]	39 (16%)	-0.81 (0.04)
MODERATE psychosocial concern was experienced [-1]	122 (49%)	
NO psychosocial concern was experienced [0]	81 (33%)	
Cannot be determined [0]	5 (2%)	
Global item: Taking into account all of the results you have just rated for this test, the genetic testing that my patient had		
Prompted better care for my patient or his/her family [2]	62 (25%)	0.53 (0.06)
May prompt better care for my patient or his/her family in the future [1]	8 (3%)	
Did not change the care provided to my patient or his/her family [0]	177 (72%)	

** Number inside bracket indicates the score that applies to the item.*

^ Items 5-7 and 9-13 are not applicable when the proband is deceased.

+ Item 1-16 has a C-GUIDE score ranges from 0 to 2; item 17 has a C-GUIDE score ranges from -2 to 0. Two probands were deceased before results disclosure.

C-GUIDE Clinician-reported Genetic testing Utility InDEx; SD standard deviation