

Supplementary Material

TITLE

Measuring the patient experience in rare disorders: benefit of pragmatic mixed-methods research in NUT carcinoma

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Supplementary Table 1: Results from cognitive debriefing of questionnaire

Item	Interpretation	Relevance	Response options	Other
QLQC30: 01 Do you have trouble doing strenuous activities, like carrying a heavy shopping bag or a suitcase?	n=1 C define “strenuous” more specifically, e.g. children don’t often lift heavy shopping bags or suitcases	n= 1 C said not relevant to child/teenager n=1 P states least important; no prob carrying or lifting	n=2 P would give different answers at different points in time; depends on when question is asked (e.g., winter vs summer)	

QLQC30: 02 Do you have any trouble taking a long walk?	n=2 P what does “a long walk” mean? Time? Distance?			
QLQC30: 03 Do you have any trouble taking a short walk outside of the house?	n=1 P what classifies a short walk?			
QLQC30: 04 Do you need to stay in bed or a chair during the day?	n= 1 P what does “you can’t walk” mean?	n=1 P not relevant r/t NUTca pertains to upper body n=1 C Most important		
QLQC30: 05 Do you need help with eating, dressing, washing yourself or using the toilet?	n= 2 C need to preface the question; before during or after Tx?	n=1 C states not relevant n=1P states least important		
QLQC30: 06 Were you limited in doing either you work or other daily activities?	n=1 C define ‘activities’ more specifically n= P parse if Tx or cancer is making Pt tired n=1 C parse answers to reflect limited due to Tx, or cancer, or COVID-19	n=1 P states not relevant; on disability n=4 C Most important is getting through the day/ADLs require help		
QLQC30: 07 Were you limited in pursuing your hobbies or other leisure time activities?	n= 1 P only because of NUTca? Or other reasons?	n=1 P states most important; can’t interact with family members as desired n=1 C states most important; 6-16	n=1 P unsure of etiology of problem	
QLQC30: 08 Were you short of breath?	n=1 P specify reason(s) for being short of breath, e.g. from playing a sport? Or from reg daily activities?	n=1 C states most important; 6-16 n=1 P states not relevant		
QLQC30: 09 Have you had pain?	n=1 P unclear on detail of pain. n=1 C Specify timeframe; now? Or in the past?	n=1 C states not relevant n=1 C states most important; 6-16		
QLQC30: 10 Did you need to rest?	n= 1 P Clarify for what reason(s) person needs rest; from exertion? Or from regular ADLs?	n=1 C states not relevant n=1 C states most important; 6-16		n=1 P overlap with QLQC30. 12 and 18 n=1 P overlap with QLQC30 18
QLQC30: 11 Have you had trouble sleeping?	n= 1 P patient not sure is inability to sleep is related to Ca or not; or drug or not	n=1 C states not relevant n=2 C states most important; 6-16 as well as another independent respondent		

QLQC30: 12 Have you felt weak?		n=1 C states most important; 6-16		n=1 P overlap with QLQC30. 10 and 18
QLQC30: 13 Have you lacked appetite?		n=2 P states not relevant n=3 C states most important; 6-16 and two other respondents		
QLQC30: 14 Have you felt nauseated?	n=1 P unclear on definition of 'nauseated'	n=1 P states not relevant n=1 C states most important; 6-16		
QLQC30: 15 Have you vomited?		n=1 P states not relevant n=1 C states most important; 6-16		
QLQC30: 16 Have you been constipated?	n=1 P include option for if you have taken painkillers	n=1 C states not relevant n=1 C states most important; 6-16		
QLQC30: 17 Have you had diarrhea?		n=2 P states not relevant n=2 C states not relevant n=1 C states least important		
QLQC30: 18 Were you tired?		n=1 P states most important		n=1 P overlap with QLQC30. 10 and 12 n=1 P overlap with QLQC30 10 n=1 P overlap with QLQC30 20
QLQC30: 19 Did pain interfere with your daily activities?	n= 1 P Patient expresses confusion about SOB and fatigue equating with pain	n=1 C states not relevant n=1 C states most important		
QLQC30: 19 Have you had difficulty in concentrating on things, like reading a newspaper or watching television?		n= 1 C states not relevant		
QLQC30: 21 Did you feel tense?		n=1 P states not relevant to self but notes that it is likely relevant to others	n=1 P states most important n=1 C states most important	
QLQC30: 22 Did you worry?		n=1 P states not relevant n=1 P states most important		n=1 C overlap with QLQC30 23

		n=4 C states most important		
QLQC30: 23 Did you feel irritable?	n= 1 C Origin of irritability should be clearer. Because of Tx? Or because you have the disease?	n=1 P states not relevant n=1 P states most important n=3 C states most important		n=1 C overlap with QLQC30 22
QLQC30: 24 Did you feel depressed?		n=1 P states not relevant n=1 P states most important n=1 C states most important		
QLQC30: 25 Have you had difficulty remembering things?		n=1 C states not relevant n=1 C states least important; attributes memory to meds for other indications		
QLQC30: 26 Has your physical condition or medical treatment interfered with your family life?		n=1 P states not relevant r/t treatments were several years ago n=1 P states most important n=1 C states most important n=1 P says COVID affects family life but not physical or medical treatments n=1 C states not relevant		n=1 C thinks 26 is "kind of a ridiculous question"; Remove n=1 C Recommend that 26, 27, 28 be reworded; then the questions will be relevant
QLQC30: 27 Has your physical condition or medical treatment interfered with your social life?		n=1 C states not relevant r/t wording n=2 C states not relevant; wording is "inappropriate", "mildly offensive" n=1 P states most important n=1 C states most important		n=1 C Recommend that 26, 27, 28 be reworded; then the questions will be relevant
QLQC30: 28 Has your physical condition or medical treatment caused you financial difficulties?		n=1 P states least important n=2 P state most important n=2 C state most important n=1 C states not relevant		n=1 C Recommend that 26, 27, 28 be reworded; then the questions will be relevant
QLQC30: 29 How would you rate your overall health during the past week?	n= 1 C it's hard to answer about overall health when person	n=2 C state most important	n=1 P thinks rating scale should be 1 to 5	

	has a rare and aggressive cancer			
QLQC30: 30 How would you rate your overall quality of life during the past week?		n=3 P state most important n=3 C state most important	n=1 P thinks rating scale should be 1 to 5	
Systemic: 31 Did you feel sleepy during the day?		n=1 P most important: notes first three questions as important n=3 C most important: energy is integral to daily living		
Systemic: 32 Have you had any difficulty carrying on with your usual activities because of getting tired easily?		n=3 C most important: energy is integral to daily living		n= 1 P Overlap w/33
Systemic: 33 Have you had a lack of energy?	n=1 P wants qualification of what tiredness means	n=1 C Irrelevant: question already asked on other surveys n=1 P most important n=4 C most important		n= 1 P Overlap w/32
Systemic: 34 Have you been dizzy?	n=1 P wants distinction between excessive sweating at night vs day n=1 C are dizzy and lightheaded the same?	n=3 P Irrelevant: just does apply n=5 C Irrelevant: "not at all"; not relevant for pt at any time n=1 P most important n=1 C most important		
Systemic: 35 Have you had fever or chills?		n=3 P Irrelevant: never experienced n=2 C Irrelevant: not observed n=1 P Least important n=2 C most important		
Systemic: 36 Have you sweated excessively?		n=3 P Irrelevant: didn't experience n=1 C Irrelevant: didn't observe n=1 C least important		
Systemic: 37 Have you had night sweats?		n=3 P Irrelevant: didn't experience n=1 P least important: n=1 C least important:		
GI: 34 Have you had cramps in your abdomen?		-n=1 caregiver found this not relevant to their experience		
H&N: 31 Have you had pain in your face?	n=1 P described head pain as from the forehead to the back of the head	n=1 P found this not relevant to their experience		n=1 P described conceptual overlap between Q31, Q32, and Q33

				n=1 P described conceptual overlap between Q31 and Q35 n=1 P and n=1 C reported this as the most important question
H&N: 32 Have you had pain in your head?				n=2 P and n=1 C reported conceptual overlap between Q32 and Q33 n=1 P reported this as the most important question
H&N: 33 Have you had headaches?	n=1 P described this as more of a sinus headache			n=2 P and n=1 C reported conceptual overlap between Q33 and Q32 n=1 C reported this as the most important question
H&N: 34 Have you had any discomfort in your neck?		n=1 P found this not relevant		n=1 P reported this as the least important question
H&N: 35 Have you had aches around your sinuses?		n=1 P and n=1 C found this not relevant		n= 3 P and n=2 C described conceptual overlap between Q35 and Q41 n= 1 P described conceptual overlap between Q35, Q32, and Q31 n=1 P reported this as the least important question
H&N: 36* Have you had nasal dryness?		n=1 C found this not relevant		n=3 P reported having this after treatment
H&N: 37* Have you had a stuffy nose?		n=2 C found this not relevant		n=1 P described conceptual overlap between Q37 and Q40 n=1 P flagged that they were having difficulty answering the question due to reconstructive surgery
H&N: 38 Have you had problems with your sense of smell?		n=1 P found this not relevant		

H&N: 39* Have you had a runny nose?		n=2 P and n=3 C found this not relevant		n=3 P reported having this problem after undergoing treatment
H&N: 40* Have you had a blocked nose?	n=1 P described blocked as ability only to mouth breathe	n=1 P and n=3 C found this not relevant		n=1 P described conceptual overlap between Q37 and Q40 n=1 P and n=1 C reported this as the most important question
H&N: 41* Have you felt pressure around your sinuses?		n=1 P found this not relevant		n= 3 P and n=2 C described conceptual overlap between Q35 and Q41 n=1 C reported this as the most important question
H&N: 42 Did you have dry mouth?				n=1 C reported this as the most important question n=8 P reported having this after undergoing treatment
H&N: 43 Have you had problems with your sense of taste?				n=1 C reported this as the most important question n=3 P reported experiencing this after undergoing treatment
H&N: 44 Did you have blurred vision?		n=3 P found this not relevant		n= 1 P described conceptual overlap between Q44 and Q45 n=2 P and n=2 C reported this as the most important question
H&N: 45 Did you have double vision?		n=3 P found this not relevant		n=1 P described conceptual overlap between Q44 and Q45 n=1 P and n=4 Cs reported this as the most important question
H&N: 47 Have your eyes been watery?		n=3 C found this not relevant		n=1 P reported experiencing this after undergoing treatment

H&N: 48* Have you had tunnel vision?		n=2 P and n=1 C found this not relevant		n=1 C reported this as the least important question
H&N: 49 Have you had any swelling in your neck?	n=1 P described this not as swelling but as deformation in the neck	n=1 P found this not relevant		n=1 C reported this as the least important question n=1 P reported completing exercises to not lose range of motion
H&N: 50* Have you had any stiffness in your neck?				n=1 C reported this as the most important question
H&N: 51 Have you had any problems chewing?				n= 1 P described conceptual overlap between Q51, Q52, Q53 n=1 C reported this as the most important question n=1 P reported this is from having dry mouth n=1 P reported this is due to pain after treatment
H&N: 52 Have you had trouble swallowing?				n= 1 P described conceptual overlap between Q51 and Q52 n=2 Ps described conceptual overlap between Q52 and Q53 n=1 P and n=2 Cs reported this as the most important question n= 3 Ps reported experiencing this after undergoing treatment
H&N: 53 Have you had any trouble drinking liquids?				n=1 P found conceptual overlap between Q53 and Q51 n=2 Ps described conceptual overlap between Q53 and Q52 n=1 C reported this as the most important question
H&N: 54				n=1 P and n=3 Cs reported this as the

Have you had problems speaking clearly?				most important question
Impacts: 31 Have you had pain while lying down?	n=1 P noted that it was not pain but just discomfort n=1 P described this as passive pain	n=1 P found this not relevant		n=1 P and n=1 C reported this as the least important question n=1 P and n=3 C reported this as the most important question n=1 P had difficulty choosing an answer because they have a difficult time quantifying their pain
Impacts: 32 Have you been limited in doing minor household repairs and maintenance (e.g. changing a light bulb or hanging up a picture)?		n=1 P and n=2 C found this not relevant		n=1 C reported conceptual overlap between Q32 and Q33
Impacts: 33 Have you been limited in doing light recreational activities (e.g. watching TV, playing cards, or reading)?	n=1 P described recreational activities as physical activities	n=1 C found this not relevant		n=1 C reported conceptual overlap between Q32 and Q33 n=1 C reported conceptual overlap between Q33 and Q34 n=1 C found it difficult to differentiate between if it was caused by cancer or the treatment
Impacts: 34 Have you felt motivated to continue with your normal hobbies and activities?	n=2 P found this question confusing as it was a positively phrased compared to the surrounding questions			n=1 C reported conceptual overlap between Q33 and Q34 n=1 P suggested phrasing it as "have you not felt motivated?" n=3 C reported this as the most important question
Impacts: 35* Have you been concerned about your ability to care for others (e.g. children, parents, spouse)?	n=1 P questioned if it was physical care or emotional support	n=1 P and n=1 C found this not relevant		n=2 C suggested adding care for a pet n=2 P and n=1 C reported this as the most important question

Lung: 31 Did you feel like you could not get enough air?		n=1 P found this not relevant		n=1 P and n=1 C reported this as the most important questions
Lung: 32 Have you coughed?				n=1 C reported this as the least important question n=1 C reported this as the most important question
Lung: 34 Did you have trouble breathing in?				n=1 C reported conceptual overlap between Q34 and Q35
Lung: 35 Did you have trouble breathing out?				n=1 C reported conceptual overlap between Q34 and Q35
Lung: 36* Have you felt pressure in your chest?				n=1 P and n=2 C reported this as the most important question
Pain: 31 Have you had aches or pains in your bones?		n=1 P and n=1 C found this not relevant		n=1 C reported this as the most important question
Pain: 32 Have you had pain in your face?	n=1 C questioned if this included the mouth	n=3 C found this not relevant		n=1 C reported this as the most important question
Pain: 33* Have you had pain in your head?		n=1 C found this not relevant		n=1 C reported this as the most important question
Pain: 34 Have you had headaches?				n=1 P reported conceptual overlap between Q34 and Q36
Pain: 35 Have you had any discomfort in your neck?		n=2 C found this not relevant		
Pain: 36* Have you had any aches around your sinuses?		n=1 P and n=3 C found this not relevant		n=1 P reported conceptual overlap between Q34 and Q36 n=1 P reported this as the most important question
Pain: 37 Have you had any pain in your back?				n=1 C reported this as the most important question
Pain: 38 Did you have abdominal pain?		n=1 C found this not relevant		n=1 C found this was the least important question
Pain: 39 Have you had pain in your chest?		n=1 P found this not relevant		n=2 C reported this as the most important question

Pain: 40 Have you had pain in your shoulder?				n=3 P reported this as the most important question
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*Denotes that item was newly developed versus from supplemental from EORTC Item Library