

Table- Questionnaire measures^a

concept				
scenario/ perspective	item	original number	question	answer categories
comprehension of issues				
	1	1	The first symptoms of Pompe disease ...	-start in infancy -can appear at any age -don't know
	2	2	For babies with classic infantile Pompe, enzyme replacement therapy is most effective when given...	-as soon as possible -before late-onset disease sets in -don't know
	3	3	A couple who each have one "incorrect" gene for alpha-glucosidase can have a baby who will later on in life develop symptoms of a later-onset type of Pompe disease.	true false don't know

4	4	Heel stick screening can, without additional testing, distinguish between classic infantile and later types of Pompe disease.	true false don't know
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acceptability of various screening outcomes

<u>classic infantile</u> <u>Pompe</u>	5	1	In this scenario, what are the most important benefits of screening in your opinion?	Open
	6	2	What are the most important disadvantages in your opinion?	open
<u>false positive</u> <i>child's</i> <i>perspective</i>	7	4	What do you think is the net effect of the screening on this child in its first year of life?	5-point bipolar scale: very harmful to very beneficial
	8	5	What do you think is the net effect of the screening on this child over its entire life?	5-point bipolar scale: very harmful to very beneficial

<i>parents' perspective</i>	9	6	If you were the parent, how much harm (for example anxiety) would you probably experience?	3-point scale: a lot of harm to little harm
<i>population perspective</i>	10	3	Is it acceptable to you if this situation occurs 60 to 100 times per year in the Netherlands?	yes
				no
<u>early detection of late-onset Pompe</u>				
<i>child's perspective</i>	11	7	What do you think is the net effect of the screening on this child in its first year of life?	5-point bipolar scale: very harmful to very beneficial
	12	8	What do you think is the net effect of the screening on this child over its entire life?	5-point bipolar scale: very harmful to very beneficial
	13	10	How much harm (for example discrimination) do you think the child would experience?	3-point scale: a lot of harm to little harm
	14	11	This child itself did not choose to be screened for a late onset disease. How do you feel about this?	I mind. I don't mind.

<i>parents' perspective</i>	15	9	If you were the parent, how much harm (for example anxiety) would you probably experience?	3-point scale: a lot of harm to little harm
<i>population perspective</i>	16	12	Is it acceptable to you if this situation occurs 3 to 5 times per year in the Netherlands?	yes no

overall acceptability of screening

screening as a public health duty	17	13	All things considered, do you think the government should offer parents of newborns screening for Pompe disease?	yes no
screening as a personal choice	18	15	If you were offered screening of your newborn child for Pompe disease, do you think you would take the offer?	probably probably not

moral reasoning					
valuation of reasons for government offer of screening	19	14	• Chance of health gain for child • Genetic knowledge gives parents new choices in further family planning. • Chance to prevent suffering • Chance for better quality of life for the child • Parental duty to gather health information on child • Other reason, namely...[space to specify]		for each reason: 3-point scale: not important to very important
decisive reason for own use of screening	20 ^b	16	6 reasons as above		forced choice of 1
valuation of reasons against government offer of screening	21	14	• Too many false positives • The child doesn't make the choice for information about possible late-onset disease • Screening adds too little to quality of life of children • An outcome of 'possibly late-onset Pompe disease' is too burdensome for a growing child • An outcome of 'possibly late-onset Pompe disease' is too burdensome for parents • Other reason, namely...[space to specify]		for each reason: 3-point scale: not important to very important

decisive reason	22 ^c	17 6 reasons as above	forced choice of 1
against own use			
of screening			

Demographics

23	21	Age	open
24	22	Gender	male
			female
25	23	Do you, or does anybody in your family have a genetic disease?	yes
			no
26	24	Do you, or does anybody in your family have Pompe disease?	yes
			no
27	25	If applicable, what is your religion affiliation?	open
28	27	What is your highest completed level of education?	8 categories + “other”
29	28	In which country were you born?	open
30	29	In which country was your mother born?	open
31	30	In which country was your father born?	open

suitability of questionnaire

32	26	If you skipped any questions [in the entire questionnaire], can you indicate why?	open
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^aThe original order of the questions is shown in Additional file 2 and in italics in column 3.

^bThis item was only to be answered by probable users of screening (item 18).

^cThis item was only to be answered by probably non-users of screening (item 18).