Supplementary information

Table S1. Importance given to requirements for responsible implementation of genome wide testing in screening

Genome-wide testing may only be used in screening if…

Topic	Requirement	All*	Prof	User
1	1. The screening participant is well informed about the screening	1.58	1.64	1.47
1	2. A pre-test counselling session with a healthcare professional with relevant expertise is a precondition for access to screening	0.33	0.64	-0.2
1	3. The government bears the responsibility for ensuring the quality of the information provided	-0.28	-0.2	-0.4
1	4. There is proper information provision about the screening and participants can request an additional pre-test counselling session with a healthcare professional with relevant expertise	1.1	1.36	0.67
1	5. Possible consequences (both positive and negative) are sufficiently explained to the screening participants	1.8	1.6	2
1	6. Possible consequences (both positive and negative) are sufficiently understood by the screening participants	1.5	1.36	1.73
2a	7. The provision of screening requires a license issued by a governmental body	0.08	0.32	-0.33
2a	8. An independent review committee has assessed that the potential benefits outweigh the disadvantages	0.2	0.44	-0.33
2a	9. The implementation meets quality criteria set by the government	0.53	0.6	0.4
2a	10. The participant can request and view the raw data (the personal genetic code that has not yet been analysed and interpreted)	-1.2	-1.4	-0.73
2a	11. The analysed data are reviewed by a recognized (genetic) laboratory specialist	0.63	0.8	0.33
2a	12. The ratio between false positive results (a result that is wrongly reported as abnormal) and false negative results (a result that is wrongly reported as normal) is aligned with the relevant form of screening according to existing rules	0.5	0.84	-0.07
2a	13. The performance meets quality criteria established by professional groups of clinical geneticists and (genetic) laboratory specialists	1.18	1.32	0.93
2a	14. The provider ensures the quality of the genome-wide testing	-1.1	-0.72	-1.6
2b	15. The chance of detecting incidental findings is reduced to a minimum	-0.08	0.16	-0.05
2b	16. Individual participants are allowed to decide for themselves on which genetic results (including incidental findings) they do or do not want to have reported back	0.25	0.04	0.6
2b	17. Professional groups of clinical geneticists and (genetic) laboratory specialists determine which genetic results (including incidental findings) may or may not be reported back to participants	-0.53	-0.04	-1.33
2b	18. The government determines which genetic results may or may not be reported back to participants	-1.9	-1.84	-2
2b	19. All findings (including incidental findings) that can be influenced by medical intervention (prevention, check-ups or treatment possible) are reported back, even if they are not part of the disorders screened for	-0.63	-0.8	-0.33
2b	20. Only genetic results related to the disorders targeted by the screening are reported back (so no reporting of incidental findings)	-0.9	-0.88	-0.93
3	21. The government ensures safe data collection, analysis and storage	-0.13	-0.6	0.67
3	22. Individual providers ensure safe data collection, analysis and storage	-0.85	-1.04	-0.53
3	23. Screening participants have been sufficiently informed about the analysis and storage of their data prior to collection of this data	0.6	0.2	1.27
3	24. Screening participants have given permission for the analysis and storage of their data prior to collection of these data	1.1	1.16	1
3	25. The collected data are destroyed after analysis	-2.5	-2.6	-2.33
3	26. Permission is requested from the person from whom the DNA originates for each possible future use of the stored data	-0.1	-0.48	0.53
4	27. Screening is made available free of charge to everyone who is eligible to participate	-0.5	-0.56	-0.4
4	28. Screening participants can invoke their right not to know at any time, meaning that they can indicate that they do not want to be informed about (certain) results	1.08	0.84	1.47
4	29. Children's right not to know is protected. This means that in the case of screening during pregnancy or in children up to 12 years of age (where the parents have the right to make the decision), only results may be provided about (the risk of) disorders for which treatment is already possible and desirable before the age of 12	0.93	0.84	1.07
4	30. Children's right to know is protected. This means that in the case of screening during pregnancy or in children up to the age of 16, the government ensures, for example through legislation and regulations, that children are informed when they reach the age of 16 about clinically relevant results of previously performed genome-wide testing	-0.05	-0.4	0.53
4	31. Participants have indicated in advance that they will inform their family members when they receive feedback on results that may be relevant to them	-1.93	-2.44	-1.07
2a	32. The cost-effectiveness of the screening offer has been demonstrated	-0.88	-0.6	-1.33
2a	33. The test offer is not at the expense of proven (cost-) effective traditional methods of screening analysis	-0.1	-0.04	-0.2
2a	34. The potential risks for participants have first been investigated in a research context	0.48	0.68	0.13
2a	35. The support for the screening offer has first been investigated in a research context	-0.2	-0.2	-0.2

Topics: 1) information and consent, 2) quality of screening (2a) and handling of incidental findings (2b), 3) privacy, responsible storage and (re-)use of data, and 4) consequences of non-participation. * The numbers provided are the average score for each requirement for all participants, the involved professionals and potential users. The score for each requirement ranges from -4 (all rate the requirement as least important) to +4 (all rate the requirement as most important).

Table S2. *Participants characteristics for the Q-sort study*

Characteristic	**Participants, n = 40 (%)**
Region	
North	3 (8%)
East	3 (8%)
South	5 (12%)
West	29 (72%)
Gender	
Female	28 (70%)
Age	
≤ 40 years	7 (18%)
41- 60 years	26 (65%)
≥ 61 years	7 (18%)
Type	
Professionals	25 (63%)
- Genetic lab specialist	4
- Clinical geneticist	3
- Genetic ELSI research experts	3
- Healthcare professionals	3
- Population screening experts	5
- Opportunistic screening experts	3
- DTC-GT experts	4
Potential users	15 (37%)
Population screening perspective	4
Opportunistic screening perspective	5
DTC-GT perspective	3
Low SES perspective	3
Experience or familiarity with screening (more than one option possible)	
Population screening	29 (18 professionals, 11 potential users)
Opportunistic screening	16 (12 professionals, 4 potential users)
DTC-GT	14 (9 professionals, 5 potential users)
No experience	4
Experience or familiarity with Genome-wide testing in screening	
Yes	30 (23 professionals, 7 potential users)
No	10 (2 professionals, 8 potential users)

DTC-GT: direct-to-consumer genetic testing, SES-socio-economic status.

Figure S1. Sorting grid for the Q-sort. The anchors are -4 least important to +4 most important. Each participant had to place one requirement in each rectangle of the grid. The sorting grids form the input for the factor analysis.

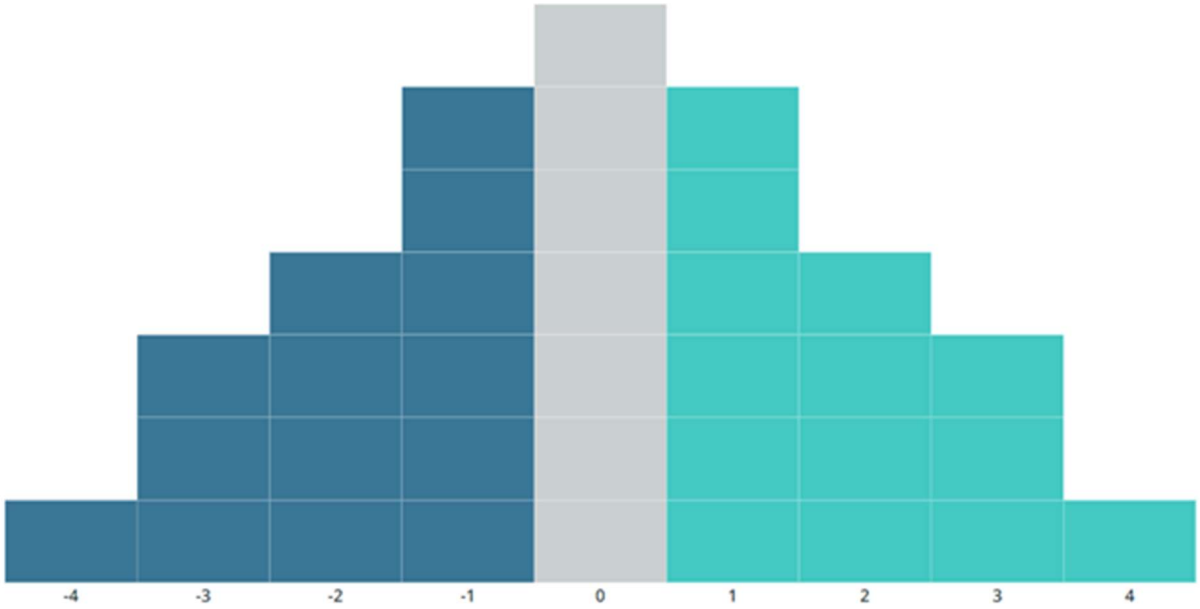

Table S3. Different perspectives towards responsible implementation of genome-wide testing in screening*

		F1*	F2*	F3*
	Eigenvalue (EV)	12.2	3.76	3.5
	Variance (%)	30.4	9.4	8.7
Genome-wide testing may only be used in screening if…				
1 - 1	the screening participant is well informed about the screening	2	4	2
2 - 1	a pre-test counselling session with a healthcare professional with relevant expertise is a precondition for access to screening	3	1	0
3 - 1	the government bears the responsibility for ensuring the quality of the information provided	2	0	-1
4 - 1	there is proper information provision about the screening and participants can request an additional pre-test counselling session with a healthcare professional with relevant expertise	3	2	1
5 - 1	possible consequences (both positive and negative) are sufficiently explained to the screening participants	1	3	4
6 - 1	possible consequences (both positive and negative) are sufficiently understood by the screening participants	1	2	3
7 - 2a	the provision of screening requires a license issued by a governmental body	4	0	-1
8 - 2a	an independent review committee has assessed that the potential benefits outweigh the disadvantages	3	-1	0
9 - 2a	the implementation meets quality criteria set by the government	1	1	1
10 - 2a	the participant can request and view the raw data (the personal genetic code that has not yet been analysed and interpreted)	-2	0	-2
11 - 2a	the analysed data are reviewed by a recognized (genetic) laboratory specialist	1	0	0
12 - 2a	the ratio between false positive results (a result that is wrongly reported as abnormal) and false negative results (a result that is wrongly reported as normal) is aligned with the relevant form of screening according to existing rules	1	0	1
13 - 2a	the performance meets quality criteria established by professional groups of clinical geneticists and (genetic) laboratory specialists	2	1	2
14 - 2a	the provider ensures the quality of the genome-wide testing	-3	-1	0
15 - 2b	the chance of detecting incidental findings is reduced to a minimum	-1	-2	3
16 - 2b	individual participants are allowed to decide for themselves which genetic results (including incidental findings) they do or do not want to have reported back	-1	3	-1
17 - 2b	professional groups of clinical geneticists and (genetic) laboratory specialists determine which genetic results (including incidental findings) may or may not be reported back to participants	0	-1	-1
18 - 2b	the government determines which genetic results may or may not be reported back to participants	-1	-3	-2
19 - 2b	all findings (including incidental findings) that can be influenced by medical intervention (prevention, check-ups or treatment possible) are reported back, even if they are not part of the disorders screened for	-2	1	-3
20 - 2b	only genetic results related to the disorders targeted by the screening are reported back (so no reporting of incidental findings)	-1	-3	0
21 - 3	the government bears the responsibility for the safe collection, analysis and storage of the data	0	0	-2
22 - 3	if providers ensure safe collection, analysis and storage of the data	-4	-1	-1
23 - 3	screening participants have been sufficiently informed about the analysis and storage of their data when collecting this data	-1	2	1
24 - 3	screening participants have given permission for the analysis and storage of their data when collecting this data	1	3	1
25 - 3	the collected data is destroyed after analysis	-3	-4	-3
26 - 3	permission is requested from the person from whom the DNA originates for each possible future use of the stored data	-2	-1	0
27 - 4	screening is made available free of charge to everyone who is eligible to participate	0	0	-3
28 - 4	screening participants can invoke their right not to know at any time, whereby they can indicate that they do not want to receive feedback on (certain) results	0	2	1
29 - 4	children's right not to know is protected. This means that in the case of screening during pregnancy or in children up to 12 years of age (where the parents have the right to make the decision), only results may be provided about (the risk of) disorders for which treatment is already possible and desirable before the age of 12	0	1	3
30 - 4	children's right to know is protected. This means that in the case of screening during pregnancy or in children up to the age of 16, the government ensures, for example through legislation and regulations, that children are informed when they reach the age of 16 about clinically relevant results of previously performed genome-wide testing	0	1	-2
31 - 4	participants have indicated in advance that they will inform their family members when they receive feedback on results that may be relevant to those family members	-3	-2	-4
32 - 2a	the cost-effectiveness of the screening offer has been demonstrated	-1	-3	-1
33 - 2a	the test offer is not at the expense of proven (cost-) effective traditional methods of screening analysis	-2	-2	2
34 - 2a	the potential risks for participants have first been investigated in a research context	2	-2	2
35 - 2a	the support for the screening offer has first been investigated in a research context	0	-1	0

Topics: 1) information and consent, 2) quality of screening (2a) and handling of incidental findings (2b), 3) privacy, responsible storage and (re-)use of data, and 4) consequences of non-participation. Importance: -4 least important to +4 most important.

* Factors were extracted and analysed to see what different perspectives the group represented. Factor analysis is the standard analysis technique in Q methodology. The sorting grids (see Appendix, Figure 1) form the input for the factor analysis. Factors were found by correlating each participant's Q-sort to determine the extent to which they had a similar configuration of the Q-sort. A three-factor solution was the optimal principal component analysis solution. After varimax rotation, 34 participants represented the three-factor solution. Six participants could not be attributed to one of these three factors.

Table S4. *Participants attributed to each perspective*

Type of participant	**Perspective 1**	**Perspective 2**	**Perspective 3**	**Not assigned**	**Total**
Professional	9	7	8	1	25
Potential user	1	6	3	5	15
Total	10	13	11	6	40