Additional file 1: Survey Questions

Demographic information:

1.   Please select the age range that best describes you

- 18-24
- 25-34
- 35-44
- 45-54
- 55-64
- 65-74
- 75-84
- 85+
- Prefer not to say

2.   Please select the gender that best describes you

- Male
- Female
- Other
- Prefer not to say

3.   Please select the ethnicity that best describes you

- White
- Asian / Asian British
- Black / Black British / African / Caribbean
- Mixed / Multiple ethnic groups
- Other
- Prefer not to say

4.   Please select the education level that best describes you

- No formal education
- Secondary (GCSE or equivalent)
- A-levels (Highers or equivalent)
- Vocational
- Undergraduate
- Postgraduate/professional
- Prefer not to say

5.   Please select the sexuality that best describes you

- ○ Heterosexual (Straight)
- ○ Homosexual (Gay or Lesbian)
- ○ Bisexual
- ○ Other
- ○ Prefer not to say

Familiarity with rare and difficult-to-diagnose diseases:

6.   Based on your knowledge before taking this survey, to what extent do you agree with the following statement? I know more about rare diseases than most people

> Strongly disagree / Disagree / Neutral / Agree / Strongly agree

7.   Based on your knowledge before taking this survey, to what extent do you agree with the following statement? I know more about difficult-to-diagnose diseases than most people

> Strongly disagree / Disagree / Neutral / Agree / Strongly agree

General sentiment towards health data sharing:

8.   To what extent do you agree with the following statement? In general, health data should be shared to improve healthcare

> Strongly disagree / Disagree / Neutral / Agree / Strongly agree

Technology in the Rare Disease Action Plan:

9.   To what extent do you agree with the following statement? The NHS should use technology to identify rare disease patients

> Strongly disagree / Disagree / Neutral / Agree / Strongly agree

10.   To what extent do you agree with the following statement? My own GP practice or hospital should use technology to identify rare disease patients

> Strongly disagree / Disagree / Neutral / Agree / Strongly agree

GP practice or hospital using technology to identify people who may have a rare disease:

11.   How likely are you to opt out of the use of your health data for research and planning, given that your GP practice or hospital can use it for this purpose?

> Very unlikely / Unlikely / Neither likely nor unlikely / Likely / Very likely

12.   If asked, how likely would you be to give your GP practice or hospital permission to use your health data for this purpose?

> Very unlikely / Unlikely / Neither likely nor unlikely / Likely / Very likely

13.   To what extent do you agree with the following statement? I would like to be informed about the use of technology to identify undiagnosed rare disease patients in my GP practice or hospital

> Strongly disagree / Disagree / Neutral / Agree / Strongly agree

14.   If your GP practice or hospital informed you that they planned to use technology to identify undiagnosed rare disease patients, which of the following methods of communication would you feel comfortable with?

- The website of your GP practice or hospital
- Posters in the GP practice or hospital
- Flyers in the GP practice or hospital
- The email newsletter of your GP practice or hospital
- A text message
- A letter sent via the post

15.   Which one of the following methods of communication do you feel would be the most appropriate?

- The website of your GP practice or hospital
- Posters in the GP practice or hospital
- Flyers in the GP practice or hospital
- The email newsletter of your GP practice or hospital
- A text message
- A letter sent via the post

The NHS using technology to identify people who may have an undiagnosed rare disease:

16.   How likely are you to opt out of the use of your health data for research and planning, given that the NHS can use it for this purpose?

> Very unlikely / Unlikely / Neither likely nor unlikely / Likely / Very likely

17.   If asked, how likely would you be to give the NHS permission to use your health data for this purpose?

> Very unlikely / Unlikely / Neither likely nor unlikely / Likely / Very likely

18.   To what extent do you agree with the following statement? I would like to be informed about the use of technology to identify undiagnosed rare disease patients by the NHS

> Strongly disagree / Disagree / Neutral / Agree / Strongly agree

19.   If the NHS informed you that they planned to use technology to identify undiagnosed rare disease patients, which of the following methods of communication would you feel comfortable with?

- ○ The NHS website
- ○ An email
- ○ A text message
- ○ A letter sent via the post

20.   Which one of the following methods of communication do you feel would be the most appropriate?

- ○ The NHS website
- ○ An email
- ○ A text message
- ○ A letter sent via the post

Opinions on being contacted further after a technology identified you as having a possible undiagnosed rare disease:

21.   If your GP practice or hospital contacted you about a potential rare disease diagnosis, how likely is it that you would feel any of the following?

- Worried that I may have a rare disease
- Grateful that my own GP practice or hospital was proactively looking out for me
- Comforted that my own GP practice or hospital is evaluating me for a rare disease
- Concerned that this had taken place without my consent

Very unlikely / Unlikely / Neither likely nor unlikely / Likely / Very likely

22.   If you think you may feel anything other than the emotions listed above, please feel free to share your thoughts here.

[Open text response]

Opinion on Mendelian and MendelScan:

23.   To what extent do you agree with the following statement? The NHS should use technology like MendelScan

Strongly disagree / Disagree / Neutral / Agree / Strongly agree

24.   To what extent do you agree with the following statement? My own GP practice or hospital should use technology like MendelScan

Strongly disagree / Disagree / Neutral / Agree / Strongly agree

25.   How likely are you to opt out of the use of your health data for research and planning, given that your GP practice or hospital can use it with MendelScan?

Very unlikely / Unlikely / Neither likely nor unlikely / Likely / Very likely

26.   If asked, how likely are you to give your GP practice or hospital permission to use MendelScan on your health data?

Very unlikely / Unlikely / Neither likely nor unlikely / Likely / Very likely

27.   To what extent do you agree with the following statement? I would like to be informed about the use of MendelScan in my GP practice or hospital

Strongly disagree / Disagree / Neutral / Agree / Strongly agree

28.   If MendelScan was used on your health data, how likely is it that you would feel worried about the following:

- The privacy and security of my health data
- How accurate the technology is
- A medical device developed by a private company has access to my health data
- The emotional distress or disruption to my life if I was identified as possibly having an undiagnosed rare disease
- If I don't have a rare disease, yet MendelScan incorrectly identified me as possibly having one, I may end up having unnecessary tests or referrals

Very unlikely / Unlikely / Neither likely nor unlikely / Likely / Very likely

29.   If you have any other concerns about MendelScan, please feel free to share them with us. We want to hear from you so that we can address your concerns and make sure that the study is as beneficial as possible for everyone involved. Please do not include information in your response that may identify you or others.

[Open text response]

30.   Do you have any other comments, positive or negative, on technologies like MendelScan that you would like to share?

[Open text response]

31.   How did you know about the survey:
- Mendelian
- The NHS
- A rare disease group
- Social media
- Other
- Prefer not to say