Oxford Medical Genetics Laboratories

User Satisfaction Survey 2016 – Next Generation Sequencing

Dear User,

Many thanks for participating in our 2016 survey about Next Generation Sequencing. We really appreciate the time you have taken to provide us with feedback.

Below is a summary of the questions posed, with a further section detailing user comments and laboratory actions.

Question 1 – Availability of NGS

In this question we asked users if there were any disease areas they would like to see an NGS panel test developed for. The breakdown of responses is as follows:

- 16 users (47%) currently refer patients for sequential Sanger tests, and would be interested in an NGS service for these disorders.
- 13 of these users supplied a comment for this question, of which 9 contained actionable requests for new panels. The following table shows these requests:

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Number requested</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neuromuscular</td>
<td>1</td>
</tr>
<tr>
<td>Endocrine</td>
<td>2</td>
</tr>
<tr>
<td>PDH deficiency and related disorders</td>
<td>2</td>
</tr>
<tr>
<td>mtDNA depletion</td>
<td>1</td>
</tr>
<tr>
<td>Epilepsy</td>
<td>2</td>
</tr>
<tr>
<td>Congenital myasthenic syndromes - the neuromuscular junction</td>
<td>1</td>
</tr>
</tbody>
</table>
Question 2 – genes included on our current panels

This question asked users about genes on existing NGS panels. The bar chart below shows the general trend across all panels.

- Overall, the majority of respondents agreed that their most relevant and key genes were on the panels, and that the panels contained the correct number of genes.

**Key to questions**

1. The panel contains the main ‘key’ genes I am interested in
2. The panel has the relevant genes on it
3. The panel has too many genes on it
4. The panel has too few genes on it

![Overall trend chart]

Question 3 – clinical priorities

This question asked respondents to rate the importance of various criteria relating to clinical priorities when considering current NGS panel tests.

- Overall, the majority of respondents stated that high percentage coverage of key genes in the panel was the most important factor. The remaining factors were given more or less equal importance.

**Key to questions**

1. Number of genes included
2. Quicker turn-around times
3. High percentage coverage of key genes
4. High percentage coverage of all genes
5. Availability of data on additional genes following initial screen
6. Low cost
Question 4 – coverage

This question reminded respondents that currently the clinical report provides the percentage coverage for all genes in the test, in the form of a table. Users were asked to rate the importance of coverage data.

- Overall the majority of respondents stated that they would like to continue seeing the detailed coverage breakdown on the report.
- The complementary question was answered as expected, with roughly the same number of replies stating that they disagreed with the statement that coverage was not important.
- There was not a strong consensus either way on whether coverage information should be stored online and looked up as required.

Key to questions

1. I would like to see a breakdown of coverage per gene, on the patient report
2. If it were possible, I would like the coverage data to be available, but I would be happy to look it up online
3. Detailed coverage information is not important to me
Question 5 – classification by number/breakdown of evidence

This question asked users to rate a number of criteria regarding the breakdown of evidence, and reminded users that we do not currently provide a classification number.

- Around one third of users stated that they either had no opinion or disagreed with the assertion that the classification was easy to discern from the report. It may be the case that this should be made clearer and/or easier to find.
- Around one third of users either had no opinion or disagreed with the notion that the classification number should be included on the report. The remaining two thirds either agreed or strongly agreed.
- 90% of those who responded indicated that they did in fact read the supporting evidence for each variant.
- 91% of respondents stated that they found this breakdown useful.

Key to questions

1. It is always clear as to which classification has been assigned to a variant
2. I would like to see the classification number stated on the report
3. Having the classification number on the report would help clarify which classification has been assigned
4. I always read through the evidence listed for each variant
5. I find the breakdown of evidence for each variant on the report useful
6. I would be happy for the classification number to REPLACE the breakdown of evidence for each variant
7. I would like to see the classification number AND evidence stated on the report
Question 6 – clinical conclusion

This question asked users to comment on both the clarity of wording and ease of locating the clinical conclusion on the report.

- The majority (90% in both cases) agreed that the clinical conclusion was clear and easy to find.

**Key to questions**

1. The wording of the clinical conclusion is clear
2. It is easy to find the clinical conclusion in the report

Question 7 – exomes

This question asked users to let us know if they currently use a diagnostic exome, and if not, whether or not they would be interested in doing so.

- Just over half currently use an exome service
- 84% would be interested in using one (this figure includes those who stated they already use an exome service).

**Key to questions**

1. Do you currently use an exome service?
2. Would you be interested in using an exome service?
Question 8 – further comments

This question provided a space for any further comments on any aspects of our NGS services. 16 users gave further feedback, which is summarised in the section below.

Summary of Comments and Actions

We received a number of very complimentary comments about our NGS services, including the design of our panels, the clarity of our clinical reports and our interpretation of variants. A number of suggestions were made for possible new NGS panels. The NGS team, together with the relevant disease teams, will be evaluating these. These could be implemented either as part of a potential upcoming exome service, or as new targeted panel tests.

A recurring comment made in both the question about coverage and in the exome section was that sufficient coverage of key/important genes was of paramount importance to our service users, and that a breakdown of coverage on a per gene basis was an important tool in understanding the report results.

We are currently investigating the possibility of introducing an exome test; a benefit for service users should be increased flexibility with regard to panel design and faster incorporation of new genes into ‘virtual’ panels. If this evaluation indicates such a test should be brought into service, we will take the strong views of our service users on coverage into account.

We received positive feedback about the clarity of our reports, with the majority of respondents feeling that our reports were clear and easy to understand. We will however continue to strive to improve the overall clarity when wording the report, in particular the clinical conclusion, to ensure it is easy to locate and understand. We will be evaluating the implications of introducing a classification number system to denote the pathogenicity of variants; however it was clear from the responses from service users that this should be in addition to a breakdown of the evidence for pathogenicity and not a replacement.