Pseudomyxoma peritonei (PMP) is a clinical syndrome characterized by gross mucinous ascites originating from a disseminated intra-peritoneal neoplasm. Although typically confined to the abdomen, mortality is high if untreated. Biomarkers, including genetic mutation profiles, may aid treatment selection and decision making. Next generation sequencing approaches such as whole-exome-sequencing (WES) can be applied to gain a broad view of the genome of these neoplasias.

**Aims**

To investigate the exome wide mutational profile of PMP in a pilot project.

**Patient samples**

All patients (A-E; Table 1) underwent debulking surgeries at the Peritoneal Malignancy Institute, Basingstoke, and tissue was archived as FFPE blocks (Figure 1).

**Methods**

FFPE blocks underwent laser capture micro-dissection to obtain cellular material of neoplastic and normal origin, from which DNA was extracted. WES was performed, aiming to generate an average depth of 60x and 100x for the normal and neoplastic material respectively. All samples had a relatively low somatic mutation burden across the exome (Table 2), and contained the frequent KRAS and GNAS mutations, with none containing TP53 mutations.

**Results**

Good quality sequence data was obtained for all samples, with an average read depth of 68x and 113x for the normal and neoplastic material respectively. All samples had a relatively low somatic mutation burden across the exome (Table 2), and contained the frequent KRAS and GNAS mutations, with none containing TP53 mutations.

**Conclusions**

We have successfully applied WES to PMP samples, achieving high purity of neoplastic cells from FFPE. LOH appears to be an important event in the pathogenesis of some PMP cases. Investigating genes present within recurrent regions of LOH in PMP would provide a powerful way to identify new genes involved in disease processes, and thus potential novel treatment targets.

**Future work**

We are currently undertaking targeted sequencing a large scale project to sequence ~400 fresh frozen PMP samples. This will allow us to combine the deep clinical phenotypes with genomic data, further developing our understanding of PMP.

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