IDENTIFICATION AND CHARACTERISATIONS OF RARE GENETIC VARIANTS (CISD2) IN A GROUP OF NHS PATIENTS BEING TREATED FOR SEVERE OBESITY. Paper details: use R software to interpret the results and construct the graphs. Methodology: - Use e a test of proportions to check whether the frequency of a particular variant in the PMMO differs from its frequency in the general population (gnomAD controls). - Check the reliability of the genotyping by visual examination of cluster plots. If the cluster plots are convincing then I will annotate these variants to assess how they might exert their effects by Combined Annotation Dependent Depletion (CADD score). - Determine what the gene does, according to the literature. Are there already disease/phenotypic associations in humans or animal models? - Looking at the gene variants in the PMMO dataset: are there mutation carriers in the PMMO? - Are the variants over- or under-represented in the PMMO compared to gnomAD controls? - Annotation of the variants – what type of mutation is? does it affect an amino acid - how? where in the protein does it occur? What is the evidence that this mutation may affect function? If time allows, repeat this analysis with related genes