Objective and methods

To determine the underlying cause of FGD by Sanger, targeted or whole exome sequencing techniques.

Figure 3: Mutations in European Countries. The most common FGD causing mutation per country varies between nationalities.

Figure 4: MC2R Mutations identified in Europeans. The most common mutations in MC2R differ between countries. S74I is the most common variant within Europe.

Figure 5: MRAP Mutations Identified in Europeans. The most prevalent mutations in MRAP differ between countries. The splice junction of coding exon 1 (c.106) is the most common mutated loci within Europe.

Conclusions

- The work has highlighted ‘mild’ presentations of several adrenal insufficiency disorders, in particular non-classical presentations of lipid congenital adrenal hyperplasia and P450 side chain cleavage enzyme deficiency with partial loss-of-function variants in STAR and CYP11A1 respectively.
- Future studies, to decipher whether causative defects are in non-coding parts of known genes, are due to copy number variation or novel genetic aetiologies will form improve genetic diagnosis of patients presenting with FGD.

C.J. Smith

c.j.smith@qmul.ac.uk