followed by 11 abnormal results recorded on chromosome 15, 10 abnormal results on chromosome 22 and abnormal results on chromosome 8.

Conclusion: Array CGH is recommended as the first line diagnostic test in patients with unexplained developmental delay, congenital abnormalities and intellectual disability due to its high diagnostic accuracy in detecting CNVs that are clinically significant.

OP3.088

The Malta NGS Project: Identifying local disease-causing variants using high throughput sequencing

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Introduction: High throughput sequencing (HTS) has revolutionised genetics research and is finding widespread application in clinical diagnostics of Mendelian disorders greatly improving on the diagnostic sensitivity of traditional approaches. The Malta NGS project was designed to take advantage of the genetic history and composition of the Maltese population to identify novel disease-causing genes and mutations in a variety of conditions and disease.

Methods: Affected singletons, trios and families were recruited following approved ethical procedures in collaboration with clinicians from Mater Dei Hospital. Blood or saliva were collected and an interviewer led questionnaire was carried out. In total, 15 conditions both rare and common are being investigated. A candidate gene approach using a custom designed capture panel of 700 genes of interest, and whole exome sequencing were used to generate HTS data. Raw data was mapped to GrCh37 as paired-end libraries using NextGENe software. An in-house data analysis pipeline was set up to shortlist potentially causative variants which were filtered against an in-house database of more than 100 HTS datasets.

Results: A number of variants which can be classified as pathogenic, likely pathogenic or influence the risk for the development of disease have been identified. Some of these are novel, not previously reported variants. A number of founder effects have also been identified.

Conclusion: HTS has been successfully used to identify causative mutations in a number of genetic conditions relevant to the Maltese population. This highlights the importance of studying the local population as many of our causative variants differ from the common northern European variants.

Disclosures: This work was carried out as part of a collaboration between the University of Malta and Mater Dei Hospital. It was supported by national funding through the R&I 2012 programme (NGS Project R&I-2012-024) administered by the Malta Council for Science and T

OP3.089

Progress report on familial hypercholesterolaemia in Malta 2018: the current situation and what needs to be done to improve it

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Introduction: FH is an autosomal dominant condition causing premature cardiovascular disease. It is under-diagnosed and undertreated worldwide-yet cost-effective treatment is available which should be started early to prevent disease progression. Expected Maltese prevalence is up to 1,700 individuals. National Premature Years of Life Lost under 65 years (PYLL-65) from cardiovascular disease is high, and FH a likely contributor. Moreover, Schedule V database has only 7 children listed as 'genetic dyslipidaemia' (expected number c.300). This represents a public health challenge to cardiologists, physicians, general practitioners and paediatricians.

Methods: A Register was established in 2017 as an observational study based on opportunistic testing, with Dutch Lipid Clinic Network phenotypic criteria for inclusion of index cases. Monitoring of progress and quality criteria is ongoing. Cascade testing uses lower LDL cut off points for family member identification.

Results: Less than 10% of potentially affected individuals have been identified; 96% of these are on statins, with 60% meeting LDL goals; most 'Definite cases' are not achieving target. One patient is on PCSK9 inhibition. Cascade testing has identified five cases to date.

Conclusion: Under 10% of affected patients have been identified on the register, with none under 18 years. Measures to raise awareness are ongoing, including presentations to various audiences. Outcomes could be improved through the formulary inclusion of ezetimibe, and PCSK9 inhibitors. Genetic testing would improve identification, facilitating the more accurate identification of children and their early management.

Disclosures: No direct funding was proviided for this study. Sanofi has funded fees for consulting and lectures, and participation in seminars related to the area.12

OP3.090

A novel mutation in the human PDHA1 gene as a cause for X-linked hereditary episodic ataxia

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Introduction: Ataxia is a disorder which presents with varying degrees of loss of control of normal bodily movements. Although acquired ataxia may be encountered,