INTRODUCTION

Targeted capture of genomic regions of interest for massively parallel sequencing allows simultaneous screening of mutations in hundreds of loci in genetically heterogeneous human diseases. It has become a widely used method in genetic testing because it leads to increased cost-effectiveness, shorter turn-around-time, higher accuracy and accessibility. Nasopharyngeal Cancer (NPC) is uncommon in many parts of the world with the age-standardized rates of about 2.2 per 100,000 but is one of the most prevalent cancers in Southeast Asia especially among the Bidayuhs and Kadazans, with its aetiology understood. Familial clustering shows that individuals with first degree family history of NPC have elevated risk of NPC with odd ratios ranged from 2 to 20 as compared to those with no family history of NPC. As genetic factors such as tumor suppressor genes that confers susceptibility to NPC remain unclear and understudied in different ethnic groups in Malaysia, we conduct Whole Exome Sequencing and Targeted Sequencing in 24 NPC cases with family history of NPC to evaluate the feasibility of targeted sequencing as well as to identify genetic factors that may lead to development of NPC.

METODOLOGY

RESULT

DISCUSSION & CONCLUSION

Genetic susceptibility to cancer is usually caused by dominantly inherited heterozygous mutations of tumour suppressor genes. However, tumour suppressor genes which confer such increased risk of NPC are understudied among various high risk groups in Malaysia. Comprehensive analysis were performed to identify potential susceptibility genes and in-depth characterization and validation of the target genes should be done to evaluate the association of such genes in pathogenesis of NPC. In our study, the targeted sequencing data demonstrates very high correlation with the WES data and our results show that both platforms are useful for the identification of potential tumour suppressor genes in NPC. It is found that in NPC, pathways related to innate immune system and DNA repair are affected. This will further enhance our understanding in the molecular basis of the disease and shed light for future prevention, diagnosis, gene therapy and/or management of NPC.

REFERENCES


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