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Mutation Profiling of Paediatric Solid Tumours in a Cohort of South African Patients

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Abstract : Background: The incidence of childhood cancer incidence is increasing gradually in low-middle income countries, such as South Africa. Globally, there is an extensive range of familial- and hereditary-cancer syndromes, where underlying germline variants increase the likelihood of developing cancer in childhood. Next-Generation Sequencing (NGS) technologies have been key in determining the occurrence and genetic contribution of germline variants to paediatric cancer development. We aimed to design and evaluate a candidate gene panel specific to inherited cancer-predisposing genes to provide a comprehensive insight into the contribution of germline variants to childhood cancer. Methods: 32 paediatric patients (aged 0-18 years) diagnosed with a malignant tumour were recruited, and biological samples were obtained. After quality control, DNA was sequenced using an ion Ampliseq 50 candidate gene panel design and Ion Torrent S5 technologies. Sequencing variants were called using Ion Torrent Suite software and were subsequently annotated using Ion Reporter and Ensembl's VEP. High priority variants were manually analysed using tools such as MutationTaster, SIFT-INDEL and VarSome. Putative identified candidates were validated via Sanger Sequencing. Results: The patients studied had a variety of cancers, the most common being nephroblastoma (13), followed by osteosarcoma (4) and astrocytoma (3). We identified 10 pathogenic / likely pathogenic variants in 10 patients, most of which were novel. Conclusions: According to the literature, we expected ~10% of our patient population to harbour pathogenic or likely pathogenic germline variants, however, we reported about 3 times (~30%) more than we expected. Majority of the identified variants are novel; this may be because this is the first study of its kind in an understudied South African population.

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