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A custom gene panel for interrogating pediatric overgrowth disorders and tumor predisposition

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My laboratory is the predominant Australian testing laboratory for pediatric overgrowth disorders associated with increased cancer risk in childhood, including Beckwith Wiedemann syndrome (BWS) and Hemi-hypertrophy (HH). Cascade testing typically involves SNP microarray, methylation analysis of imprinting centers on 11p15.5 and CDKN1C (P57) mutation screening. Rare point mutations in NSD1, NLRP2, DNMT1 and ZFP57 have been described in BWS and like disorders as well as deletions and insertions within the 11p15.5 imprinting centers IC1 (H19/IGF2) and IC2 (KCNQ1OT1/CDKN1C). Tumor risk is increased in most genetic and epigenetic subtypes of BWS and HH however degree of risk and tumor type varies between groups. Parents of affected children are often understandably anxious to know the recurrence risk for these conditions and as the number of childhood cancer survivors' increases, the possibility for transmission of a causative mutation is becoming an increasingly important issue. To improve our capacity to detect predisposing mutations in BWS, HH and in the pediatric tumors that have been described in these conditions, we have designed a gene panel comprising 37 genes as well as inter-genic regions spanning imprinting centers on 11p15.5 and 11p13. We have used the Haloplex target enrichment system with sequences run on an Illumina MiSeq. We have performed pilot testing to show that the panel has clinical utility and demonstrates excellent sequence coverage of the 11p imprinting centers. Analysis of results to date has revealed novel mutations including OCT-4 binding site disruption in IC1 and sub-regions of homozygosity.

Biography

Elizabeth Algar was awarded a PhD from Griffith University Australia in 1989 and since then she has been working in the broad area of cancer genetics with a specific focus on pediatric cancer. She has led research teams at several universities and research institutes in Australia including the University of Queensland and University of Melbourne. She has authored 60 publications and holds several awards for her work. She has been a panel member of several national grant bodies and is regularly called upon to review journal articles and grant applications. She presently holds the positions of Principal Scientist at Monash Health and Assoc. Professor at Monash University and the Hudson Institute.

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