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Abstract Topic:- Clinical Genetics

Abstract Title:- Cockyane syndrome cases in Indian population and estimating if c.4063-1G>C is a common variant.

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Aims:- Cockayne syndrome is a phenotypically heterogeneous rare disorder that presents with a spectrum of clinical features including cachectic dwarfism, wrinkled skin, subcutaneous fat loss, beaked nose, and stooped posture. The disease is inherited in autosomal recessive pattern and occurs as a result of disease-causing homozygous or compound heterozygous variations in ERCC6.

Methods:- Exome sequencing was performed on Illumina sequencing platform and raw data obtained was analysed in-house. The reads were aligned to human reference genome 37/hg19 using Burrows Wheeler Aligner (BWA-MEM). Variant calling was done using PICARD tools and GATK software package (https://gatk.broadinstitute.org/hc/en-us) and annotated using Annovar (http://wannovar.wglab.org) and an in-house bioinformatics pipeline. Run of homozygosity (ROH) was analysed using Automap v1.2 aligned with human reference GRCh37.

Results:- All the 6 patients reported with ERCC6 variation in this cohort had a common c.4063-1G>C variant. Among these 6 cases, one patient (patient 3) had compound heterozygous variant involving the common c.4063-1G>C variant and another c.1357C>T variant in ERCC6 gene. Homozygosity mapping was performed using AutoMap tool, identified 1.36 Mb homozygous region in chromosome 10 (Chr10:50186415-51549314) common in all the samples.

Conclusions:- This suggests that the variant c.4063-1G>C in ERCC6 gene is likely to be a common variant in Indian population rather than a hotspot variant.

Keywords:- Cockayne syndrome; ERCC6 gene; Exome Sequencing, Homozygosity Mapping.