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Drawing on the sum of human genomics experiences to identify disease causing mutations

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A key challenge in genome interpretation and precision medicine is the lack of an extensive, high quality, ethnically-diverse collection of human genomes as a reference set. A prospective disease-causing variant that appears to be "rare" based on publicly available sequence may in fact be a polymorphism in an ethnic population under-represented in public databases. Resources such as the Exome Variant Server, the 1000 Genomes Project, and the Exome Aggregation Consortium have been immensely valuable to the community, and Kaviar combines such datasets into integrated allele frequencies, but public databases have not been funded to provide broad and deep ethnic representation. QIAGEN's Ingenuity Variant Analysis™ genome interpretation solution has been used to interpret hundreds of thousands of ethnically diverse human sequencing samples.

However, these NGS datasets are private and most are never publicly released. The Allele Frequency Community (http://www.allelefrequencycommunity.org) has been formed to address this interpretation need. The AFC consist of over 100k of exome/genome variants sets representing human subjects from over 100 countries. This ethnically diverse dataset has been shown in internal benchmarking studies to generate a 43% average reduction in false positive rates in causal variant identification. Combining the experience of over 100,000s of genomes analyzed and access to expert-curated scientific literature, we have been able to solve genetic cases at a rate of roughly 87%. In the future we envision more examples of genetic samples 'recycled' for studies other than their original purpose furthering the research endeavors of many more scientists.

Optimization and comparison of different methods for RNA isolation for next generation sequencing from *Elettaria cardamomum*

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High quality RNA isolation is crucial for obtaining meaningful results during small RNA and transcriptome analysis using next generation sequencing. Due to the presence of several primary as well as secondary metabolites, no standard method of RNA isolation is applicable for all plants. The polysaccharides and polyphenols are prominent in various tissues of cardamom (*Elettaria cardamomum Maton*) which critically hinder the RNA extraction protocols, hence methods using TRIzol, TRIzol with sodium sulphite, RNeasy Plant Mini Kit, PureLink RNA Kit, CTAB, CTAB with PureLink RNA Kit, RNeasy Plant Mini with CTAB, miRNeasy Kit, miRNeasy with CTAB were attempted for obtaining intact RNA from cardamom leaf, stem, flowers, flower buds and young fruits. Our results shown that polysaccharide and polyphenol hindrances are greater in PureLink RNA Kit and TRIzol methods and the amount of RNA generated from all tissues were unsatisfactory. RNA isolated from modified CTAB method yielded RNA with high quantity and good quality as evidenced from A260/280 and A260/230 ratios but the integrity of total RNA isolated was not reliable. The RNeasy Plant Mini Kit and miRNeasy Kit extracted RNA with good purity and high yield but the secondary metabolites present in cardamom interfered the isolation process by lowering the A260/230 ratio. RNeasy Plant Mini Kit with CTAB and miRNeasy Kit with CTAB methods obtained RNA with good purity and quantity as evidenced from A260/280 and A260/230 ratios and BioAnalyzer RIN (RNA integrity number) values. The total RNA from these two methods was found amenable for next generation sequencing.