

Department of Genetics
University of Delhi South Campus

New Delhi- 110021

Limited Tender Enquiry for Exome Sequencing

Sealed tender is invited from national and international firms/institutes with experience and expertise for **exome sequencing of 20-60 Human samples with and without data analysis** to be carried out as per specifications below. The sequencing cost and data analysis cost should be mentioned separately in the tender. The tender with technical details may be sent to the under signed latest by 5pm on September 21, 2015.

1) Exome sequencing:

- Exome sequencing should be performed using the Illumina HiSeq 2000/2500
- Exome enrichment should be performed using Agilent SureSelectV5+UTR (post capture) enrichment kit
- Each sample should be sequenced to at least 50-fold (50X) mean target depth or 100X raw target depth, with 101-bp paired-end sequencing method
- For each sample >95% target regions should captured and sequenced with 10X coverage
- Minimum 8GB cleaned data/sample (adaptor and low quality sequence/reads removed) should be delivered. The proportion of clean data and raw data should be 90% or above.
- >90% of total data should be >Q30 Phred score
- Summary of complete sequencing runs should be provided.

Bioinformatic analysis of exome data

- 1) **Read quality check and data filtering:** Detailed quality report before and after trimming should be provided. It should include base quality and sequence quality score distribution details, average base content and GC distribution in the reads, PCR amplification details, check for over-represented sequences, adapter trimming details, read length details (percentage of read length distribution), parameters used for trimming and names and details of software used and other details of trimming/removal of low quality sequence/reads etc.
- 2) **Read alignment:** The paired end reads should be aligned to reference genome GRCh37/hg19. Alignment should be done using standard tools such as BWA.

Details of parameters used for alignment and software details, read alignment statistics and quality metrics for each sample should be provided.

- 3) **Details of target depth and raw depth:** Percentage of target regions captured in each sample and details of not captured regions of the target region covered by the probe and coverage depth details should be provided.
- 4) **Variant calling:** Variant calling (SNPs and INDELs) should be performed using standard procedures of GATK or Samtools. Summary reports of variants called from each sample should be provided, number of SNPs, INDELs, homozygous and heterozygous variants and transition to transversion ratio, read depth and quality distribution of identified variants in each sample should be provided. Variant calling should be done individually and together (all samples together to produce single VCF file for all samples). Variant filtration should be done using standard procedures and parameters and also provide details of procedures and parameters used for the filtering.
- 5) **Variant annotation:** Complete annotation of the variants obtained above using contemporary tools and their summary statistics should be provided. Also the names of software and parameters used for the annotation should be provided.

Sample Details: 20-60 samples will be sent to the firm.

Data delivery deadline: Sample DNA QC report should be submitted within 5 days after receiving the sample. Raw FastQ file, Clean FastQ file and other quality and parameter files mentioned above (at 1) should be delivered within 30 days after sample QC completed. BAM file, filtered and unfiltered VCF files (individual sample's VCF and all samples together) and other parameters and quality files mentioned above (at 2,3,4,5) should be delivered within 50 days after sample qc check is completed.

Other conditions

- Firm/Institute should submit evidence of prior experience for whole exome/genome sequencing
- List of clients/institutes where firm successfully completed whole exome/genome sequencing projects in last one year should be submitted

- Firm should provide evidence of technical and experience in this area for completion of this work.

Payment: Payment will be made only after the receipt of the complete data (with above criteria) in accordance with above mentioned requirements/criteria. **If data does not fulfill the above detailed criteria, the data will not be accepted and payment will not be made. If timely delivery of data is not done, penalty will be applied. In case of late delivery of data 1% of total cost will be deducted for delay of every day.**

If firm/institute does not follow the above mentioned detailed criteria the institute/Lab has the full right to cancel the agreement with the firm/institute at any time. In that case no payment would be made.

The selection of the firm for the above mentioned service will be based on lowest quoted price, evidence of expertise in the field and technical specifications mentioned in the tender.

Per sample cost for the exome sequencing of 20 to 30, 31 to 40, 41 to 50 and 51 to 60 samples with and without bioinformatics analysis may be provided.

Quotations may be sent to

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On or before September 21, 2015

Forwarded

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