STORE SECTION (D.O.)
ALL INDIA INSTITUTE OF MEDICAL SCIENCES
ANSARI NAGAR, NEW DELHI - 110029

Ref. No. XX-63/SO(DO)/EOI-Pead./2026-27/M&E **Dated: 14.09.2026**

CORRIGENDUM

It is informed that the tender bearing no. XX-63/SO(DO)/EOI-Pead./2026-27/M&E and CPP tender I.D. No. 2026_AIMSD_920773_1, Expression of Interest for Diagnostic Genomic Testing services for OPD Patients., Subsequent to pre-bid meeting, the technical Specification are revised on the feedback received from potential vendor & dates of tender has been extended, as per TSEC recommendation and detail given below:

Sl. No.	DETAIL	EXISTING	AMENDED/ EXTENDED
1.	Bid submission END Date	14.09.2026	28.09.2026
2.	Bid Opening Date	15.09.2026	29.09.2026
3.	Technical Specification	Enclosed revised technical specification. **Annexure-I**	

All other terms & condition of the tender are same.

14/9/2026

(MANOHAR ARYA)
Sr. Store Officer (DO)

ANNEXURE-I

Technical Specifications for diagnostic genomic tests

EXOME AND GENOME SEQUENCING

Background:

Whole Exome Sequencing (WES), is a comprehensive genomic diagnostic test that interrogates the protein-coding regions of the genome to identify disease-causing variants. It is recommended for individuals with suspected monogenic disorders, heterogeneous phenotypes, and unsolved cases after first-tier testing. Whole Genome Sequencing (WGS) is a comprehensive genomic diagnostic test that interrogates the entire nuclear genome, enabling detection of single nucleotide variants, small insertions/deletions, copy number variants, structural variants, and selected non-coding and regulatory region variants. WGS is increasingly recommended for complex, unsolved, or critically ill cases where prior testing (CMA, exome sequencing) is inconclusive.

This tender seeks to empanel/select qualified laboratories or service providers capable of delivering high-quality, clinically validated exome and genome sequencing services with robust interpretation and reporting.These may include either proband-only sequencing or trio sequencing (including proband and both parents).

Specification for sequencing data

The selected bidder shall provide end-to-end WES services, including:

- Sample collection guidelines and logistics
- DNA extraction (if applicable)
- Library preparation, sequencing, and bioinformatics analysis
- Clinical variant interpretation and reporting
- Turnaround time (TAT) assurance
- Reanalysis and post-test support

Sample Types and Requirements

- Peripheral blood (EDTA): 2–3 mL per individual
- Saliva / buccal swab/ Dried blood spots (if offered, with validation data)

- Prenatal samples (if offered):
    - Amniotic fluid: 10–15 mL
    - Chorionic villus sample (CVS): as per standard guidelines
- DNA quality and quantity requirements to be specified

Test Configuration

The bidder must specify available configurations:

- Proband-only WES or WGS
- Duo WES or WGS (proband + one parent)
- Trio WES or WGS (proband + both parents)
- Rapid/STAT WES or WGS

Short-read compliant technology*

Test specifications

Test Name	Whole Exome Sequencing (solo)	Whole Exome Sequencing (trio)	Whole Genome Sequencing (Short read) (solo)	Whole Genome Sequencing (Short read) (trio)	*Whole Genome Sequencing (Long read) (Solo)	*Whole Genome Sequencing (long read) (trio)
Genes Covered	>20000 genes	>20000 genes	Complete genome	Complete genome	Complete genome	Complete genome
Sequencing Platform	NGS platform utilizing short read sequencing-by-synthesis or equivalent high-accuracy chemistry	NGS platform utilizing short read sequencing-by-synthesis or equivalent high-accuracy chemistry	NGS platform utilizing short read sequencing-by-synthesis or equivalent high-accuracy chemistry	NGS platform utilizing short read sequencing-by-synthesis or equivalent high-accuracy chemistry	NGS platform utilizing true single molecule long read sequencing covering entire genome, full length transcripts & difficult areas with precise assemblies & variant calls.	NGS platform utilizing true single molecule long read sequencing covering entire genome, full length transcripts & difficult areas with precise assemblies & variant calls.

Kit Specification	Name of the kit used	Library prep must utilize a high-efficiency Target Enrichment /Hybridization-capture based protocol (enzymatic or mechanical fragmentation).	Library prep must utilize a high-efficiency Target Enrichment /Hybridization-capture based protocol (enzymatic or mechanical fragmentation).	Library prep must utilize a high-efficiency Target Enrichment /Hybridization-capture based protocol (enzymatic or mechanical fragmentation).	Library prep must utilize a high-efficiency Target Enrichment /Hybridization-capture based protocol (enzymatic or mechanical fragmentation).	Amplification free approach to retain epigenetic signals with no PCR artifacts or errors	Amplification free approach to retain epigenetic signals with no PCR artifacts or errors
	Vertical coverage (Depth)	>80X	>80X	>35-45X	35-40X	>20X with average insert size of at least 13-30Kb	>20X with average insert size of at least 13-30Kb
	Horizontal coverage	>97% coverage	>97% coverage	>97% coverage	>97% coverage	95 – 99%	95 – 99%

Raw data requirements	Fastq files	Fastq files	Fastq files	Fastq files	Fastq files	Fastq files
	BAM file	BAM file	BAM file	BAM file	BAM file	BAM file
	Raw vcf file	Raw vcf file	Raw vcf file	Raw vcf file	Raw vcf file	Raw vcf file
	Annotated vcf file	Annotated vcf file	Annotated vcf file	Annotated vcf file	Annotated vcf file	Annotated vcf file
	Coverage	Coverage	Coverage	Coverage	Coverage	Coverage

All specification, target size & chemistry performance requirements mentioned herein are defined purely by operational & clinical quality metrics. Offers meeting equivalent or superior specification on alternative platforms/ chemistries will be evaluated provided they meet the defined quality benchmarks (Q30)more than 90% raw reads, Coveragethresholds, uniform capture performance & TAT.

Capture and Coverage

- Clinical-grade exome capture kit to be specified
- ≥95% of targeted exonic regions covered at ≥20× depth
- Mean target coverage: ≥80–100× (to be specified)
- Coverage metrics to be included in the report

Sequencing Technology

- Massively Parallel Sequencing (NGS)
- Paired-end sequencing (minimum 100 bp reads) in WES and Paired-end sequencing (≥150 bp reads preferred)for WGS

Detectable Variant Types

The assay must reliably detect:

- Single nucleotide variants (SNVs)
- Small insertions and deletions (indels)
- Copy number variants (exome-based CNV calling – if offered, specify limits)
- Homozygosity/ROH (if SNP data available)

- For WGS (short read)
    - Structural variants (deletions, duplications, inversions, translocations) with stated limits
    - Repeat expansions (if bioinformatically supported, specify loci)
    - Regions of homozygosity (ROH) and uniparental disomy (UPD)
    - Mitochondrial genome variants (preferred; specify depth)

WGS Long read-

Data should be able to interpret all genomic aberrations including SNPs, inDels, SVs, repeat expansion, and methylation for each sample. Individual files for each variation must be provided.

- Limitations must be clearly stated, including:
- Low-level mosaicism detection limits
- Structural variants and repeat expansions (WGS)

Analytical Performance and Validation

Bidders must provide:

- Platform and pipeline validation data
- Sensitivity and specificity for SNVs, indels, CNVs, and SVs
- Minimum variant allele fraction detectable
- Reproducibility and QC performance metrics

Bioinformatics Pipeline

- Validated alignment, variant calling, and annotation pipeline
- Use of up-to-date reference genome (GRCh38 preferred; specify if GRCh37)
- Annotation using multiple databases (ClinVar, gnomAD, OMIM, HGMD or equivalent)
- Phenotype-driven analysis using HPO terms (preferred)

Variant Interpretation and Classification

- Variant classification according to ACMG/AMP guidelines (latest version)

- Structural variant interpretation using ACMG/ClinGen recommendations
- Clear categorization: Pathogenic, Likely Pathogenic, VUS, Likely Benign, Benign
- Gene-disease validity based on ClinGen or equivalent resources

Reporting Requirements

The WES/WGS report must include:

- Patient identifiers and detailed clinical phenotype
- Test configuration (proband/duo/tri/rapid)
- Sequencing metrics and coverage summary
- Identified clinically relevant variants with:
    - Gene name and transcript
    - Genomic coordinates and HGVS nomenclature
    - Zygosity and inheritance pattern
    - Clinical interpretation and phenotype correlation
- Secondary/incidental findings policy (ACMG SF list – opt-in/opt-out)
- Recommendations for confirmatory testing (e.g., Sanger validation)and family studies
- Clear limitations of the test
- Authorized signatory (Medical Geneticist / MD / PhD Genetics)

Turnaround Time (TAT)

- Proband-only exome: 3-4 weeks
- Proband-only Genome: 6-8 weeks
- Trio exome: 3-4 weeks
- Trio Genome-6-8 weeks
- Prenatal/Expedited/STAT exome(for time bound situations)-10-14 days
- Rapid exome/genome (critically sick patients)- 5-7 days
- At least 99% sample must fulfil the above mentioned TAT

Quality and Accreditation

The laboratory must:

- Be accredited by NABL (India) / CAP for NGS testing
- Participate in External Quality Assurance Schemes (EQAS) for NGS
- Maintain documented SOPs, IQC, and audit trails

Ethical, Legal, and Regulatory Compliance

- Written informed consent covering scope, limitations, and secondary findings
- Compliance with national biomedical ethics guidelines
- Compliance with data protection and privacy laws
- For prenatal testing, compliance with PCPNDT Act

Data Management, Storage, and Reanalysis

- Secure data storage for FASTQ/BAM/VCF files
- Data sharing only with authorized stakeholders
- Defined data retention period
- Complimentary reanalysis within 12–24 months
- Policy for data sharing and patient access

Logistics and Sample Handling

- Defined sample acceptance/rejection criteria
- Cold-chain and transport requirements (if applicable)
- Mechanism for repeat analysis in case of QC failure or some other technical reason

Evaluation criteria

Bids will be evaluated based on:

- Technical capability and coverage quality
- Interpretation depth and clinical reporting quality
- Accreditation and quality systems
- Turnaround time
- Cost-effectiveness and experience

Expected numbers of NGS samples per month

Exome- 250-300

Genome- 50-100

Specification for Chromosomal Microarray

Chromosomal Microarray Analysis (CMA) is a first-tier diagnostic test for the detection of copy number variations (CNVs), including microdeletions and microduplications, across the genome. It is widely recommended for individuals with developmental delay, intellectual disability, autism spectrum disorder, multiple congenital anomalies, and selected prenatal indications. This tender aims to identify and empanel qualified laboratories/service providers capable of delivering high-quality, reliable, and clinically interpretable CMA services in compliance with national and international standards.

The selected bidder shall provide end-to-end CMA services, including:

- Sample collection requirements and guidelines
- Laboratory processing and analysis
- Bioinformatics analysis and CNV interpretation
- Clinical reporting
- Turnaround time (TAT) assurance
- Post-test clarification and quality assurance support

Sample Types and Requirements

- Peripheral blood (EDTA): 2–3 mL (postnatal)
- Prenatal samples (if offered):
    - Amniotic fluid: 10–15 mL
    - Chorionic villus sample (CVS): as per standard guidelines
- Products of conception: fresh tissue or DNA (with clear acceptance criteria)

Test Name	Microarray
Total Probes	>7,00,000
CNV Probes	> 5,00,000
SNP Probes	>2,00,000
Detection Limit	
a) Gain	Till 200Kb
b) Loss	Till 100 Kb
c) Mosaicism	Till >15%
d) LOH analysis	-
Raw data requirements	Cel files/raw files
	Library QC Report
Turn around time	7-10 days

The assay must be capable of detecting:

- Copy number gains and losses
- Microdeletion and microduplication syndromes
- Aneuploidies and large chromosomal imbalances
- Mosaicism (with stated detection limits)
- Regions of homozygosity / consanguinity (for SNP-based arrays)

Balanced rearrangements (e.g., inversions, translocations) shall be clearly stated as limitations.

Logistics and Sample Handling

- Clear sample acceptance and rejection criteria
- Defined transport and storage conditions
- Mechanism for repeat analysis in case of QC failure

Data Management, Data provision and Confidentiality

Secure Laboratory Information Management System (LIMS)

The final data should be made available on a hard disc drive as well as accessible on cloud for a period of two years after completion of sequencing.

Files should be compatible with third-party software.

Data sharing only with authorized stakeholders

Expected numbers of samples per month
CMA-30-50

Non-invasive prenatal screening for aneuploidies

Non-Invasive Prenatal Screening (NIPS), also referred to as cell-free fetal DNA (cfDNA) testing, is a screening test for common fetal aneuploidies using maternal blood.
The purpose of this tender is to empanel/select qualified laboratories or service providers capable of delivering high-quality, reliable, ethical, and clinically validated NIPS services in accordance with national and international standards
The selected bidder shall provide end-to-end NIPS services, including:

- Pre-analytical logistics (sample collection kits, transport)
- Laboratory testing and bioinformatics analysis
- Clinical interpretation and reporting
- Turnaround time (TAT) assurance
- Post-test support and quality assurance

Technical specifications
Sample Requirements

- Maternal peripheral blood: 8–10 mL

- Blood collection tubes validated for cfDNA stabilization (e.g., Streck or equivalent)
- Minimum fetal fraction threshold to be specified (e.g., ≥4%)

Conditions to be Screened (Minimum Requirements)

The assay must, at a minimum, screen for:

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)

Optional (clearly specified if offered):

- Sex chromosome aneuploidies (45,X; 47,XXY; 47,XXX; 47,XYY)

Laboratory Methodology

- Massively Parallel Sequencing (MPS) or validated equivalent technology
- Whole-genome or targeted sequencing approach (to be specified)
- Robust bioinformatics pipeline with validated algorithms

Performance Characteristics

Bidders must provide peer-reviewed or validation data demonstrating:

- Sensitivity ≥99% for Trisomy 21
- Specificity ≥99% for Trisomy 21
- Clearly stated sensitivity/specificity for Trisomy 18 and 13
- Reported no-call rate and redraw policy

Reporting Requirements

Reports must include:

- Patient and sample identifiers
- Gestational age at sampling
- Fetal fraction (mandatory)
- Risk classification (High risk / Low risk / No-call)
- Clear statement that NIPS is a screening, not diagnostic, test
- Recommendation for confirmatory invasive testing for high-risk results
- Authorized signatory (MD/PhD in Genetics or equivalent)

Turnaround Time (TAT)

- Maximum TAT: 7–10 working days from sample receipt

- Faster TAT (if available) should be specified separately

The laboratory must:

- Be accredited by NABL (India) / CAP / ISO 15189 or equivalent
- Participate in external quality assurance programs (EQAS) for cfDNA/NIPS
- Maintain internal quality controls and documented SOPs

Ethical, Legal, and Regulatory Compliance

- **Strict compliance with the PCPNDT Act (India), including:**
  - No disclosure of fetal sex
  - Appropriate documentation and declarations
- Compliance with data protection and patient confidentiality norms
- Informed consent templates to be provided

Expected Numbers: 40-50 per month

Evaluation Metrics for Bidders

The following criteria of the bidders be assessed:

The vendor should have an accredited lab in India

- Technical compliance and validation data
- Clinical reporting quality
- Turnaround time
- Cost-effectiveness
- Accreditation and quality systems -Lab should be NABL accredited/ISO certified
- Reporting as per standard guidelines (ACMG/AMP)
- Experience and track record

The testing laboratory must possess at least 5 years of experience in clinical sample processing. Sequenced data must originate strictly from the bidder's own wet-lab facility, with no third-party outsourcing or sub-contracting permitted.

Specification for GCMS analysis of suspected In born error of metabolism

Background:

This is to invite and establish service level agreement of a qualified, NABL-accredited analytical laboratories , provide comprehensive GC-MS–based biochemical metabolic testing for the clinical diagnosis of suspected inborn errors of metabolism (IEM).A laboratory with a capability of delivering high sensitivity , specificity analysis and data for more than 198 metabolites across multiple IEM categories with rapid turn around ,robust quality assurance and provision for emergency reporting

Scope: vendor shall provide end-to-end services for GC-MS analysis of biological samples (primarily urine, with optional plasma/DBS if offered) for suspected IEM including

SampleTypes: Urine sample,blood/DBS in case of infants and New Born

Sample condition : Fasting /Anytime specific to metabolic condition

Analyte Coverage:

- Amino acidopathies,Organicacidemias,TCA cycle intermediates,Mitochondrial disorders, Fatty acid oxidation defects, Peroxisomal disorders, Purine and pyrimidine disorders,Sugar and sugar alcohol disorders, Pipecolic acid and related markers

Services Required:

- Supply of all sample collection accessories: sterile urine vials, filter papers for newborns/infants, gloves, TRF (test requisition forms), transport boxes, and instructions.
- Complete analytical process from sample receipt to final report issuance (no subcontracting of diagnostic tests).
- Quantitative reporting in **mmol/mol creatinine** and **multiples of median (MoM)** with age/condition-specific reference ranges.
- Online report access and delivery to requesting lab within defined TAT.
- Participation in internal and external quality assurance (EQA/PT) programs.
- **Emergency/urgent reporting within 48 hours for critical cases, with fee exemption for poor patients as per agreed terms.**
- Interpretation support flagging of abnormal patterns suggestive of specific IMDs
- Report should be delivered in PDF form along with raw data files e.g..mzML, or other compatible format

Specification for GCMS Methodology: bidder must specify exactly the model configuration and details of the instruments on which testing is being offered. This must include information on ancillary instruments used for sample preparation, softwares .

1. Sensitivity and specificity ≥99% for detection of organic acidemias, amino acidopathies, Fatty acid oxidation defects and sugar alcohol disorders (supported by validation data or published evidence).
2. Full scan and SIM scan. Automated SIM setup and synchronous SIM/scan operation.
3. Latest original licensed library metabolomics such as NIST and Wiley library etc

Turnaround Time (TAT):

- Routine: within 3days from sample receipt.
- Urgent: within 48 hours from sample receipt (define surcharge if applicable).

Data Retention: Minimum retention period of [e.g., 2 years] for re-analysis or audit, under defined storage conditions

Expected numbers of samples per month

GCMS- 75-100

High resolution HLA Typing technical specs

(i) High resolution HLA typing using Next Generation Sequencing platform e.gIllumina Novaseq X Plus or illuminaor equivalent

(ii) Coverage: High coverage of all the major HLA Gene regions. Should offer a flexibility of full Gene /Partial coverage for above locus HLA Type 1 & Type 2 (HLA A, HLAB,HLA C, HLA DRB1,HLA DPB1, HLA DQB1)upto 4 fields.

(iii) Facility for isolation of genomic DNA from peripheral blood samples

(iv) NGS with 2x150 bp reads.Library preparation should be done using GenDx Kit.

(v) Should offer a flexibility of Multiplex or Singleplex based amplification strategy(06 or 11 loci) .

(vi) Complete bioinformatics data analysis. Should include the latest IMGT Database.

(vii) No extra cost should be charged from the PI for re-run the HLA typing if generated data doesn't match the desired quality

(viii) The complete scope of work should be done by the vendor within India

(ix) The vendor should provide a facility certificate to testify the possession of an in-house Next Generation Sequencing platform explicitly in the name of the Company.

(x) The vendor should have demonstrated scientific capability for the successful completion of undertaken genomics projects, as documented by the vendor having research publications in peer reviewed journals for at least 5 genomics study projects. An appropriate web link to support this claim should accompany the proposal.

(xi) The vendor should have a CAP Accredited laboratory in India.

(xii) Bidder must be providing regular HLA typing service for human disease diagnosis and must submit documentary proof for the same.