

Invitation of quotation  
for  
Outsourcing of investigations  
for genetic sequencing  
At

**All India Institute of Medical Sciences, Jodhpur**

Inquiry No. : AIIMS/Admin/RES/143(II)/2025

Inquiry Issue Date : 08 May 2025

Last Date of Submission : 14 May 2025 at 03:00 PM.



**All India Institute of Medical Sciences, Jodhpur**

Basni Phase - II, Jodhpur – 342005, Rajasthan

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**Invitation of quotation for Outsourcing of investigations  
for genetic sequencing at AIIMS Jodhpur**

Sealed Quotations are hereby invited by the undersigned on behalf of the Executive Director, AIIMS Jodhpur for Outsourcing of investigations for genetic sequencing for the Institute as per terms & conditions mentioned below. The filled quotations along with all the required document must reach in the office of the undersigned on or before 14/08/2025 03.00 PM. The Envelope containing the quotation would please be sealed and super scribed as under:-

**“QUOTATION FOR Outsourcing of investigations  
for genetic sequencing AGAINST INQUIRY NO.  
AIIMS/Admin/RES/143(II)/2024” DUE ON 14/08/2025 03.00 PM”**

**1. Terms & Conditions:**

- A) The quotations received after this deadline & unsealed shall not be entertained under any circumstances whatsoever. In case of postal delay this Institute will not be responsible. **The offer Submitted Fax/Email shall not be considered and no correspondence will be entertained in this matter.**
- B) Quotations must be in the enclosed prescribed Performa on the letter head of the firm duly signed by the Proprietor/ Partner/Director or their authorized representative, In case of signing of quotation by the authorized representative letter of authorization must be attached with the quotation. Quotation must be dropped in “**Quotation Box**” located in Administration Block of AIIMS, Jodhpur.
- C) Rates must be quoted in **Indian rupees** and as per the format specified taxes extra if any must be written separately.
- D) Rates must be quoted FOR basis (including Freight charges, Insurance, installation etc.)
- E) No overwriting or cutting is permitted in the rate. If found, the quotation shall be summarily rejected.
- F) The rates quoted must be valid for 60 days minimum from the date of opening of the quotation and silence of any tendered on this issue shall be treated as agreed with this condition.
- G) Becoming L1 will not be the criteria for awarding of purchase order unless the rates are reasonable & justified.
- H) RTGS/NEFT details need to be furnished by the supplier with the quotation on the letter head of supplier/firm/agency.
- I) The firm/agency may satisfy the following conditions and attach self-attested copy of the same with the quotation:
  - Firm shall be registered with the Government of Rajasthan / Central Government.
  - The firm shall have valid GST/Other taxes and IT PAN.
  - **The firm should not be black listed by any Govt. Agency/Dept.**
- J) Quotations qualified by such vague and indefinite expressions such as “subject to prior confirmation”, “subject to immediate acceptance” etc. will be treated as vague offers and rejected accordingly. Any conditional quotation shall be rejected summarily.

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- K) **Delivery Period** – within 15 days from Purchase order.
- L) **Liquidated Damage:** - If the supplier fails to deliver the material on or before the stipulated date, then a penalty at the rate of 0.5 % per week of the total order value shall be levied subject to maximum of 10% of the total order value.
- M) **Payment Terms:** Payment will be only after satisfactorily delivery / commissioning of all material and after inspection by the AIIMS Jodhpur.
- N) **Disputes:** -In the event of any dispute or disagreement arising between the contractors and any other department of AIIMS Jodhpur with regards to the interpretation of “Terms & Conditions” of this inquiry, the same shall be referred to the Executive Director, AIIMS Jodhpur whose decision will be final and binding upon the contractor.
- O) AIIMS, Jodhpur reserves the right to increase or decrease quantity and / or amount of work. Decision of Quantity of material for Executive Director, AIIMS, Jodhpur will be final in this regard.
- P) AIIMS, Jodhpur reserves the right to reject any quotation or part or the whole of inviting quotation process without assigning any reason. Decision of the Executive Director, AIIMS, Jodhpur will be final in this regard.
- Q) The near relatives of employees of AIIMS, Jodhpur are prohibited from participation in this tender. The near relative for this purpose are defined as: (a) Members of a Hindu undivided Family. (b) Their spouses (c) The one related to the other in the manner as father, son(s), Son's wife (daughter-in-law), daughter(s) and daughter's husband (sons-in-law) brother (s) and brother's wife, sister(s) and sister's husband, brother(s)-in-law.

### 2. Special Terms & Conditions:

- A) **Bidder must quote the product as per specification provided in Annexure 1.**
- B) **Catalog must be attached with quotation for technical evaluation.**
- C) **The Bidder must submit the GSTIN Registration**
- D) **The supplier may be asked to arranging demonstration of their equipment for which rates have been quoted, to the AIIMS Jodhpur, if required. The expenditure incurred for demonstrating the items will be borne by the supplier.**
- E) **After successful award of the order, if/whether the Bidder stated that they will not be able to supply the items for any particular reason, Executive Director AIIMS, Jodhpur reserves the right to ban such firm.**

**Dean (Research)**

Encl.: Annexure 1 (Specification)  
Annexure 2 (Format of price bid)

**Annexure 1**

| S.No. | Outsourcing of investigations | Pack Size  | Qty     |
|-------|-------------------------------|------------|---------|
| 1.    | for genetic sequencing        | 10 per set | Upto 40 |

**Specifications for Genetic Sequencing required \*****Specifications required:****Reporting of any detected gene or a copy number variation****Capture kit:** Illumina**Platform:** Illumina sequencing platform**Depth of coverage:** 100x, and more than 99% coverage at 50x**Confirmation included:** Sanger sequencing to confirm the detected variant from NGS OR a Chromosomal Microarray if a Copy Number variant is detected.**Genes to be covered:** Annexure 1**Requisites:**

1. Access to raw data file for all patients.
2. Submission of a sample raw data file for technical evaluation at user end.

The cost of quoted should be inclusive of taxes and the cost of raw file and confirmatory tests (Sanger/CMA)

The samples be analysed and released in batch of 10 at a time.

| S No. | Disorder   | Gene/location        | Protein   | Inheritance |
|-------|--|----------------------|---|-------------|
| 1     | Type 1 Antenatal Bartter syndrome  | SLC12A1(15q15-q21.1) | NKCC2   | -           |
| 2     | Type 2, aBS  | KCNJ1(11q24)         | ROMK1   | -           |
| 3     | Bartter Syndrome   | CaSR(3q 13.3-q21)    | Calcium sensing receptor  | AD          |
| 4     | Type 4 Abs, with sensorineural deafness  | BSND CLCNKA-CLCNKB   | Baiitin   | -           |
| 5     | Familial hypomagne semia, hypercalciuria and nephroncalcinosis                               | CLDN16,3q28          | Paracellin-1  | AR          |
| 6     | Familial hypomagne semia, hypercalciuria and nephrocalcinosis with severe ocular involvement | CLDN 19, 1p34        | Claudin-19  | AR          |
| 7     | Familial hypomagne semia with Hypocalciuria  | TRPM6, 9q22          | TRPM6   | AR          |
| 8     | Hypercalciuria/Calcium oxalate nephrolithiasis   | ADCY10/SAC           | Adenylate cyclase I0  | AD          |
| 9     | Primary Hyperoxaluria type1  | AGXT                 | Alanine glyoxylate aminotransferase                                     | AR          |
| 10    | Adenine phosphoribosyl transferase deficiency urolithiasis (DHA stones)                      | APRT                 | Adenine phosphoribosyl transferase                                      | AR          |
| 11    | d RTA  | ATP6VOA4             | ATP ase H <sup>+</sup> transporting, lysosomal V0subunit a4             | AR          |
| 12    | d RTA with deafness  | ATP6V 1B1            | ATP ase H <sup>+</sup> transporting, lysosomal 56/58 kDa, V 1 subunit B | AR          |
| 13    | Osteopetrosis with distal RTA  | CA2                  | Carbonic anhydrase 2  | AR          |
| 14    | Dent's Disease   | CLCN5                | Chloride channel voltage sensitive 5                                    | XR          |

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|           |   |          |   |       |
|-----------|---|----------|---|-------|
| <b>15</b> | 1,25-OH D24 hydroxylase deficiency, infantile Hypercalcemia                                 | CYP24A1  | Cytochrome P450, family 24, subfamily A, polypeptide 1  | AR    |
| <b>16</b> | Enamel renal syndrome, amelogenesis imperfect 20 and Nephrocalcinosis                       | FAM20A   | Family with sequence similarity 20  | AR    |
| <b>17</b> | Primary Hyperoxaluria type 2  | GRHPR    | Glyoxylate reductase/hydroxypyruvate reductase  | XR    |
| <b>18</b> | MODY+ Fanconi syndrome+ nephrocalcinosis  | HNF4A    | Hepatocyte nuclear factor 4, alpha  | AD    |
| <b>19</b> | Primary hyperoxaluria type 3  | HOGA1    | 4Hydroxy-2-oxoglutarate aldolase 1  | AR    |
| <b>20</b> | Kelley-Seegmiller syndrome, partial HPRT deficiency, HPRT-related gout                      | HPRT1    | Hypoxanthine phosphoribosyltransferase 1  | XR    |
| <b>21</b> | Lowe syndrome/Dent disease 2  | OCRL     | Oculocerebral renal syndrome of Lowe  | XR    |
| <b>22</b> | Renal Hypouricemia, RHUC1   | SLC22A12 | Solute carrier Family 22, Member 12   | AR/AD |
| <b>23</b> | Renal Hypouricemia, RHUC2   | SLC2A9   | Solute carrier Family 2, Member 9   | AR/AD |
| <b>24</b> | Nephrolithiasis/Calcium Oxalate   | SLC26A1  | Solute carrier family 26, member 1  | AR    |
| <b>25</b> | Calcium oxalate stones, high oxalate absorption   | SLC26A6  | Solute carrier family 26, member 6  | AR/AD |
| <b>26</b> | Hypophosphatemia nephrolithiasis/osteoporosis - 1, NPHLOP1/Fanconi renal tubular syndrome 2 | SLC34A1  | Solute carrier Family 34, Member 1  | AR/AD |
| <b>27</b> | Hypophosphatemia rickets with hypercalciuria  | SLC34A3  | Solute carrier Family 34, Member 3  | AR    |
| <b>28</b> | Cystinuria, type A  | SLC3A1   | Solute carrier Family 3 (Cystine, dibasic and neutral amino acid transporters, activator of cysteine, dibasic and neutral amino acid transport), Member 1 | AR    |
| <b>29</b> | Primary distal renal tubular acidosis, dominant/ recessive                                  | SLC4A1   | Solute carrier Family Member 3  | AR/AD |
| <b>30</b> | Cystinuria, type B  | SLC7A9   | Solute carrier Family 7(glycoprotein associated amino acid transporter light chain, bovine system), Member 9  | AD/AR |
| <b>31</b> | Hypophosphatemic Nephrolithiasis/ osteoporosis-2, NPHLOP2                                   | SLC9A3R1 | Solute carrier Family 9, subfamily A, (NHE3, cation proton antiporter Member 3  | AD    |
| <b>32</b> | Idiopathic hypercalciuria   | VDR      | Vitamin D (1,25-dihydroxyvitamin D3) receptor   | AD    |

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|    |                    |     |                        |    |
|----|--------------------|-----|------------------------|----|
| 33 | Xanthinuria Type 1 | XDH | Xanthine dehydrogenase | AR |
|----|--------------------|-----|------------------------|----|

|    | Gene           | Coded protein   | SNP, variant   |
|----|----------------|---|--|
| 1  | <i>AQP1</i>    | Aquaporin 1   | rs1000597; rs12669187  |
| 2  | <i>ALPL</i>    | Alkaline phosphatase  | rs1256328  |
| 3  | <i>CLDN14</i>  | Csludin 14  | rs219780; rs219778; rs199565725  |
| 4  | <i>TRPV5</i>   | Trnsient receptor potential calcium permeable cation channel V5 | p.L530A  |
| 5  | <i>TRPV6</i>   | Transient recptor potential cation calcium permable channel V6  | rs4987657; rs4987667; rs4987682  |
| 6  | <i>SPP1</i>    | Osteopontin   | 144 and 145; T-593A; rs491126616; rs17524488; c.240T>C; c.708C>T; rs11439060 |
| 7  | <i>CASR</i>    | Calcium sensing receptor  | rs1042636; rs1801725; E1011Q; rs6776158; rs1501899; rs7627468                |
| 8  | <i>ORA11</i>   | Calcium release activated calcium modulator                     | ra12313273   |
| 9  | <i>VDR</i>     | Vitamin D receptor  | rs2228570; rs1544410   |
| 10 | <i>KL</i>      | Klotho  | G395A; rs3752472   |
| 11 | <i>NHERF1</i>  | Sodium-hydrogen antiporter 3 (regulator 1)                      | L110V; R153Q; E225K  |
| 12 | <i>FGF23</i>   | Fibroblast growth factor 23                                     | rs7955866  |
| 13 | <i>CALCR</i>   | Calcitonin receptor   | 3'UTR+18C>T; rs 72570683; rs3214144  |
| 14 | <i>SLC13A2</i> | Na/dicarboxylate cotransporter-1                                | 1550V  |
| 15 | <i>F2</i>      | Prothrombin   | rs5896   |
| 16 | <i>IL-RN</i>   | Interleukin 1 receptor antagonist                               | Intronic promoter region   |
|    | <i>VNTR</i>    |   |  |
| 17 | <i>PON1</i>    | Paraononase-1   | rs854560   |
| 18 | <i>CARD8</i>   | Caspase recruitment domain family member 8                      | rs2043211  |
| 19 | <i>UGT1A1</i>  | UDP glucuronosyltransferase family 1 member A1                  | rs10929303; rs1042640; rs8330  |
| 20 | <i>SLC34A1</i> | Phosphate carrier NPT2a   | rs11746443; rs12654812   |

## SNP's proposed to be detected

**VDR gene:** 1544410, 731236, 2228570, 7975323, 10735810

**CaSR Gene:** 1042636, 1801726, 1801725, 7652589, 6776158, 1501899, 7627468

CLDN 14: 219780, 219778

**SLC13A2-** 1550V, 11567842

**SLC34A3-** 199690076, 762710288

**SLC34A1-** 56235845, rs 11746443, 12654812

**TRPV5** –rs 4252402, p.L530 A

| VDR            | Ca             | Klotho         | Osteopontin | Urokinase       |
|----------------|----------------|----------------|-------------|-----------------|
| 1544410        | <b>1042636</b> | <b>1207568</b> | 1126616     | 4065            |
| 731236         | <b>1801726</b> | 577912         | 17524488    | 2227564         |
| <b>2228570</b> | <b>1801725</b> | 3752472        | 2728127     | 2074725         |
| 7975232        | 17251221       | 650439         | 11739060    |                 |
|                | 1501899        | 564481         | 4754        | Sodium channels |

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|  |         |         |          |                  |
|--|---------|---------|----------|------------------|
|  | 7652589 | 9536314 | 9138     | SLC13A2-1550V    |
|  | 7648044 | 1207568 | 28357094 | 11567842         |
|  | 6776158 |         | 1126772  | Calcium channels |
|  | 7648041 |         | 2853744  | TRPV5            |
|  | 10190   |         | 11730582 |                  |
|  | 7627468 |         | 11439060 |                  |
|  | 1048213 |         |          |                  |

**Note:- The supplier may be asked to arranging demonstration of their material for which rates have been quoted, to the Administrative Block, AIIMS Jodhpur, if required. The expenditure incurred for demonstrating the items will be borne by the supplier.**

**On the letterhead of firm]**

**ANNEXURE “2”**  
**PRICE BIDFORM**

To,

**Dean (Research),**  
AIIMS, Jodhpur.

Dear Sir,

1. I/We ..... Submitted the quotation for Enquiry No. **“QUOTATION FOR Outsourcing of investigations for genetic sequencing AT AIIMS, Jodhpur AGAINST THE INQUIRY NO. AIIMS/Admin/RES/143(II)/2024”** due on 14/08/2025 03.00 PM for Outsourcing of investigations for genetic sequencing at AIIMS Jodhpur”.

2. I/We thoroughly examined, understood and accepted terms & conditions given in the enquiry document, failing which my quotation will be rejected out rightly.

3. I/We hereby offer to supply at the following rates.

| S. No. | Outsourcing of investigations | Specification      | Qty     | Price/Unit Exclusive of GST (INR) | GST/ Other Taxes | Price/ Unit Inclusive of GST (INR) | Total |
|--------|-------------------------------|--------------------|---------|-----------------------------------|------------------|------------------------------------|-------|
| 1.     | for genetic sequencing        | As mentioned above | Upto 40 |                                   |                  |                                    |       |

**Note:-**

- The Bidder must quote only single Make & Model.**
- The Bidder must submit the GSTIN Registration and PAN Card self-attested copy with the quotation otherwise quotation will be REJECTED.**
- The bidder must quoted their quotation only in above said format on the letter of firm otherwise quotation will be REJECTED.
- Catalog must be attached with quotation for technical evaluation.
- The Bidder may be asked to submit the sample of quoted make for technical evaluation at Medical College, Third Floor, AIIMS Jodhpur at the time of submission of their quotation.**

Date\_\_\_\_\_

Place \_\_\_\_\_

(Name)\_\_\_\_\_

Name of Firm/Company/Agency\_\_\_\_\_

GSTIN No.: \_\_\_\_\_

Bank Name:- \_\_\_\_\_

Bank Account No.: \_\_\_\_\_

IFSC Code:- \_\_\_\_\_

Branch Name: \_\_\_\_\_

Phone No.\_\_\_\_\_

Email:\_\_\_\_\_

(Signature of Authorized Person) \_\_\_\_\_

Seal:\_\_\_\_\_