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RAIKAMI
24 HOUR
* Medical & Impaired Mgmt. CHEN
* General & Operative
* PH: 011-237
* GET WELL - SC



Molecular Genetics
Institute of Medical Genetics & Genomics
Phone: 91-11-42251992; 42251993
Email: dnasgrh@gmail.com, sgrhgenetic@yahoo.in

2023-December

Hosp Reg No: 3292159

Request Received: 09-12-2023

NGS data ID: 23H790-Abbas [CDFD]

Referred by: Dr. Ratna Dua Puri (SGRH, New Delhi)

Molecular Lab No: 23H790

Data Type: Fastq files

NGS Lab ID: 23_215

Reporting Date: 16-03-2024

Genetics Referenc: G23-5820

Name: Abbas Hussain

DOB: 08-04-2023

Gender: Male

Clinical Indication: Abbas Hussain, 8 months old baby presented with the chief clinical complaints of failure to gain height and weight with normal development. On examination, mild frontal bossing, right fifth finger clinidactyly, smooth philtrum and thin upper lip were noted. No cafe-au-lait spots or limb asymmetry.
Clinical Impression: ? 3M Syndrome, Mulibrey nanism.

Consanguinity: No.

Clinical history: No.

Test Requested:

Exome data analysis. Data received from CDFD [23H790-Abbas]

Results:

No clinically significant "Pathogenic" or "Likely Pathogenic" single nucleotide variants/Indels identified matching with the described phenotype.

Variant Details:

| Gene Variant | Location | Zygoty | Disease | Disease Inheritance | Variant Classification (ACMG) |
|---|---|--------------|--------------------------------|---------------------|-----------------------------------|
| OBSL1:c.4043 G>A [NM_015311.3] | Exon 12 p.Arg1348His Non-synonymous Variant | Heterozygous | 3-M syndrome 2 | Autosomal Recessive | Variant of uncertain significance |

No Second sequencing variant identified in **OBSL1** gene.

Other genes [CUL7,CCDC8] associated with 3-M syndrome were also screened for sequence level variants (SNV/Indels).

Recommendations:

- # Please correlate the results with other laboratory and clinical findings.
- # Genetic Counselling is strongly recommended to explain and discuss the results.

Govt. of India

छुट्टी की पर्ची Discharge-Slip
कलावती सरन बाल अस्पताल
Kalawati Saran Children's Hospital

बंगला साहिब मार्ग, नई दिल्ली-110001
Bangla Sahib Marg, New Delhi-110001
दूरभाष / Tel. No. : 23344160, 23344162-65

युनिट Unit U2 C2 सी.आर. नं. C.R. No. 4511

नाम Name : ABBAS

आयु Age : 10 M लिंग Sex: MALE

पता Address : Mohalla Bazar Razzak Amroha U.P.

भर्ती की तारीख : 17/02/24 छुट्टी की तारीख : _____
Date of Admission Date of Discharge

निदान
Final Diagnosis : Pneumonia & Severe Perimembranous Ventricular Septal defect (PM-VSD)
& Severe Pulmonary Arterial Hypertension & Severe Anemia & Severe
Acute Malnutrition & Global Developmental delay

Anthropometry MVAL-10.1
Wt. at Admission 5.8kg (-4.81) Wt. at Discharge _____
Height/Length 63.2 (-4.81) Head Circumference 40cm (-4.48)
Nutritional Status WFL-(-3.20)

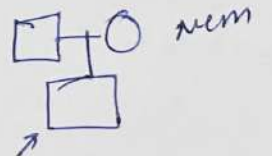
Immunisation

BCG

Pentga/DPT/OPV B1 B2

Hep. B

Measles / MMR / Typhoid





**SRI SATHYA SAI SANJEEVANI
INTERNATIONAL CENTRE FOR CHILD HEART CARE & RESEARCH**

📍 Palwal, Haryana - 121102

Totally Free Of Cost Pediatric Cardiac Care

OPD NO. : 022400/261

NAME : Abbas / 11M

DoB : 08/04/23 GENDER : ... M ... DATE : 21/03/24

CONSULTANT :



H-2008-0017
Since June 16, 2008



Sir Ganga Ram Hospital

Please note:

- Although all precautions are taken during Molecular Genetic testing the currently available data indicate that the technical error rate for all types of Molecular DNA analysis is approximately 2%.
It is important that all clinicians or persons requesting Molecular Genetic diagnostic tests are aware of these data before acting upon these results.
- The results assume that all patient information provided is correct.

Analysed by

Samarth Kulshrestha
Bioinformatics Analyst

Reviewed By:

Dr. Pallavi Sinha
DrNB Fellow

Authorised by

Dr. Ratna D Puri
Sr. Consultant, Chairperson

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Appendix: ACMG Classification, Methodology

PTO

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SRI SATHYA SAI SANJEEVANI
INTERNATIONAL CENTRE FOR
CHILD HEART CARE & RESEARCH

Sri Sathya Sai Sanjeevani International Centre For Child Heart Care & Research
Bhagola, NH-2, Delhi-Mathura Road, Palwal Dst, Haryana-121102
Telephone : +918010119000

Normal Coronaries
Left arch, No PDA/CoA
Normal Ventricular function
Severe PAH

Final Diagnosis:

Large Subaortic VSD with inlet extension BD shunt, Additional tiny apical VSD(2 shunts) BD shunt, Bilateral SVC, Severe PAH

Child Heart Care Team Decision:

Needs VSD closure. To meet surgical team. Follow up with local pediatrician for routine care, Family counselled.

Prescription Notes:

Syp Furoped 0.5ml BD
Tab Aldactone 6.25mg BD
Tonoferon drop 5 drop BD
Vitamin D3 drop 1ml OD


Advice:

IE Prophylaxis
Avoid dehydration
Vaccination as per schedule

Category:

1A

Dr. Paramvir Singh
Senior Consultant


Dr. Adhi Arya
Consultant

Dr. Anuj Sharma
Consultant

In view of long waitlist & urgency of surgery kindly seek elsewhere

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WE CARE LIKE A MOTHER

NANHI MUSKAN FOUNDATION

सेवा में
नन्ही मुस्कान फाउंडेशन
58/13 चिरगा दिल्ली,
मैन रोड नई दिल्ली - 110017.

महोदय,

का श्री. सिविनीय निवेदन यह है कि मेरा बेटा 10 माह का है, मेरे बेटे के दिमाग में छेद है दिन में छेद की वजह से इसका आँस लेने में बहुत परेशानी हो रही है।

अतः इसका इलाज बहुत महंगा हो रहा है। इसका इलाज करने में पूर्ण रूप से असमर्थ हूँ, कृपया करके मेरी मदद करें।

99
बेटे का नाम - अजय

आपकी प्राथमिक
अच्छे की माँ

श्री. राज

दिनांक

31/03/24

