Web.add : www.cancercaretrust.org
E.id : contact@cancercaretrust.org

CANCER CARE TRUST

PATIENT APPLICATION FORM

PATIENT'S DETAILS

NAME:	**CHINTU PASWAN**
FATHER NAME:	**PREMJEET KUMAR**
DATE OF BIRTH / AGE:	**13th July 2017 / 5 Years**
SEX:	**MALE**
ADDRESS:	**MITTHABEL, GORAKHPUR, UTTAR PRADESH-273203**
DISEASE:	**LEUKOCYTE ADHESION DEFICIENCY(LAD 1) BLOOD CANCER**
HOSPITAL DEPARTMENT TRATMENT TREATMENT COST	**AIIMS** **MEDICAL ONCOLOGY** **BONE MARROW TRANSPLANT** **Rs. 12 LAKHS**

For CANCER CARE TRUST

Authorized Sign

Authorised Signatory

Parent's Sign

डा. बी. आर. अम्बेडकर संस्थान रोटरी कैंसर अस्पताल
Dr. B.R. Ambedkar Institute Rotary Cancer Hospital
अ.भा.आ.सं. अस्पताल/A.I.I.M.S. HOSPITAL

OPR-6

बहिरंग रोगी विभाग/Out Patient Department

अस्पताल के अन्दर धुम्रपान मना है।/SMOKING PROHIBITED IN HOSPITAL PREMISES

एकक/Unit Prof SB/DP

विभाग/Dept. Medonco

IRCH No. 274087

ब०रो०वि० पंजीकृत सं०/O.P.D. Regn. No. 10344228

नाम/Name	पिता/पुत्र/पत्नी/पति/पुत्री F/S/W/H/D of	लिंग Sex	आयु Age	जन्म तिथि/Date of Birth
Chintu		M	4y	

निदान/Diagnosis LAD-1 / HSCT w/u (Funds pendi

दिनांक/Date	उपचार/Treatment
26/08/22	~~Adv:~~ - Paronychia. -(1) To J/u c Prof S K Kakra Si for continuing follow-up -(2) Syp. Cimid (100 mg/5 ml) 10 खाने के बाद 5 ml — 5 ml — 5 ml X 1 -(3) Syp. Cefixime (50 mg/5 ml) खाने के बाद 2 ml — 2 ml X 10da -(4) DSA - #LA-Class I & II antibodies for Donor — Anny, Preenjit -(5) GFR-DTPA scan as dated
Wt = 11kg	

(6) F/U – 23/09/22 – CBC/LFT/KFT + Blood group

(7) Arrange funds.

(151)

डा. बी. आर. अम्बेडकर संस्थान रोटरी कैंसर अस्पताल
Dr. B.R. A[illegible]lospital
अ.भा[illegible]AL
अस्पताल के [illegible]MISES

OPR-6

D Ticket

DR. B.R.A. IRCH,AIIMS,NEW DELHI
IRCH No. 27408[illegible] Reg.Date-12/05/2022
Clinic Paediatric Medical Oncology Clinic Clinic No. 2022/[illegible]74
Deptt. MEDICAL ONCOLOGY
General
UHID-103442288
नाम
Name CHINTU
S/O- PREMJIT Sex/Age M /4Y
Room 5 (Shift Mor[illegible]g)
Address JAIN CLY T 160 UTTAM NAGAR , DELHI, INDIA

एकक/Unit DR. S.B
विभाग/Dept. DR. D.P
नाम/Name
No. DDMOC-6374
जन्म तिथि/Date of Birth

निदान/Diagnosis LAD-1 / for HSCT

CTP-250622005 103442288
CHINTUPASWAN

दिनांक/Date	उपचार/Treatment
	Review 27/5/22 Meerk Did not come on 27/5/22
1/6/22	Seen by Dr Deepani Dr HLA typing – child & parents. Review ~~24/6/22~~ 16/6/22 Meerk
16/6/22	Review on 24/06/22 c HLA typing report (outside) Meerk

Parents want to plan next pregnancy
Genetic opinion – Tue/Fri Old RAK OPD

Me

24/6/22 LAD-1 /for haploidentical transplant:

1. Arrange funds.
2. HLA typing
3. Blood donation 3① - 2यू ए वी – Main Blood Bank emergency
4. CBC/RFT/LFT/Viral markers.
5. Echo.
6. GFR
7. CMV-IgG & IgM
8. Dental clearance – Dental block - दांते दांते विभाग
9. Blood group.
10. Review 26/8/22 ℂ reports.

QR Code for report verification

Case ID	: 2200120667	Sample Type	: EDTA PERIPHERAL BLOOD
Name	: - CHINTU PASWAN (CCT)	Date & Time Collected	: 29-Jun-2022 11:00 AM
Sex/Age	: Male/4 Years	Date & Time Received	: 02-Jul-2022 04:32 PM
Bill. Loc.	: AIIMS 1, Delhi (C/o Santosh)	Date & Time Reported	: 27-Jul-2022 11:39 AM
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Indication	:		

Whole Exome Sequencing on the Illumina NGS Platform.

Clinical Indication:

Leukocyte adhesion defect type-1

Positive

(Clinically significant variants detected related to the clinical phenotype)

Advise: Targeted mutation analysis of reporter variant in parents.

Key Finding: 01

Gene & Transcript	Location	Variant	Zygosity	Inheritance	Clinical Significance
ITGB2 NM_000211.5	Intron 8	c.994-1G>C (Splice Acceptor Variant)	Homozygous	Autosomal Recessive	Pathogenic

**Genetic test results are reported based on the recommendations of American College of Medical Genetics*

Phenotype	1. Leukocyte adhesion deficiency (AR-116920)

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✧ REFERENCE LAB IN MOLECULAR GENETIC DISEASES ✧

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www.sngenelab.com

CONDITIONS OF REPORTING

- It is presumed that the specimen belongs to the patient named or identified in the test request form.
- In case of collected specimen(s), which are referred to S. N. Genelab, Surat from a referral centre, it is presumed that patient demographics are verified and confirmed at the point of generation of the said specimen(s).
- A test requested might not be performed for the following reasons:
  - Specimen quantity insufficient (inadequate collection / spillage in transit)
  - Specimen quality unacceptable. (Collected in improper container)
- A test requested might yield "INVALID RESULTS" for various technical reasons and this response will appear against the test name followed by a detailed comment at the end of the report.
- If the collection date was not stated in the Test Requisition Form, the same will not be printed on the report.
- Test parameters marked by asterisks are excluded from the "scope" of NABL accredited tests.
- Some tests are referred to other laboratories to provide a wider test menu to patients.
- Tests are performed as per the test schedule given in the test listing. In unforeseen circumstances (non-availability of kits, instrument breakdown & natural calamities) tests may not be reported as per schedule. S. N. Genelab will ensure that the delay is minimized.
- This report is not valid for medico-legal purposes.
- Neither S. N. Genelab nor its Doctors and technical staff assume any liability, responsibility for any loss or damage of any nature whatsoever that may be incurred or suffered by any person as a result of use of the report.
- Isolated laboratory investigations may not confirm the final diagnosis of a disease. They help in arriving at a diagnosis in conjunction with clinical presentation and other related investigations.
- PCR is a technique used to amplify the number of copies of a specific region of DNA, in order to produce enough DNA to be adequately tested. Using specific primers, mutations in β- globin gene are detected by ARMS PCR. Currently available data indicate that the technical error rate for all types of DNA analysis is approximately 0.5%.
- Karyotyping is a standard cytogenetic technique that analyses metaphase chromosome spreads obtained from cultured cells. All genetic abnormalities like single gene / polygenic disorders, microdeletions, subtle rearrangements and low grade mosaicism cannot be ruled out by karyotyping.
- FISH is a molecular diagnostic tool for a rapid and precise identification of chromosomal aberrations (aneuploidies, microdeletions, translocations etc.) using specific commercially available DNA probes. However, only probe-specific defects get identified. Very small microdeletions, point mutations or other genetic etiologies cannot be detected by FISH.
- The sex of the fetus will not be revealed as per the Prenatal diagnostic Techniques (Regulation and Prevention of Misuse) Act 1994.
- Any query from the referring doctor pertaining to this report shall be directed to S. N. Genelab, Surat.

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Interpretation:

The submitted sample shows homozygous variant in intron 8 of gene ITGB2. A "G" to "C" substitution was detected at nucleotide position 994-1. This variant mutates a splice-acceptor sequence, but is predicted to preserve the reading frame, resulting in in-frame exon skipping.

Variant predictions:

The variant c.994-1G>C has been previously classified as Likely pathogenic in ClinVar (Variation ID 1339025 as of 2022-06-02) with respect to Leukocyte adhesion deficiency 1. The c.994-1G>C variant is a loss of function variant in the gene ITGB2, which is intolerant of Loss of Function variants

Population Variant Frequency:

The c.994-1G>C variant is observed in 1/30,782 (0.0032%) alleles from individuals of gnomAD South Asian background in gnomAD All. This variant occurs in no individuals in a homozygous genotype state. The c.994-1G>C variant is novel (not in any individuals) in 1kG All.

Gene Summary:

This gene encodes an integrin beta chain, which combines with multiple different alpha chains to form different integrin heterodimers. Integrins are integral cell-surface proteins that participate in cell adhesion as well as cell-surface mediated signalling. The encoded protein plays an important role in immune response and defects

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in this gene cause leukocyte adhesion deficiency. Alternative splicing results in multiple transcript variants.

ADDITIONAL FINDINGS: NO VARIANT OF UNCERTAIN SIGNIFICANCE (VUS) DETECTED

Genes analyzed:

ACD, ACP5, ADA, ADA2, ADAR, AICDA, AIRE, AK2, AP3B1, ARPC1B, ATM, ATP6AP1, B2M, BACH2, BLM, BLNK, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CCBE1, CD19, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CDCA7, CEBPE, CFD, CFH, CFI, CFP, CHD7, CIITA, CLPB, COPA, CORO1A, CSF2RA, CSF2RB, CSF3R, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1B, DCLRE1C, DKC1, DNAJC21, DNASE2, DNMT3B, DOCK2, DOCK8, ELANE, EPG5, ERCC6L2, EXTL3, F12, FADD, FAS, FASLG, FAT4, FERMT3, FOXN1, FOXP3, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, HAX1, HELLS, HPS1, HPS4, HPS6, HTRA2, ICOS, IFIH1, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKBKG, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RG, IL36RN, IL7R, INO80, IRAK4, IRF8, ISG15, ITCH, ITGB2, ITK, JAGN1, JAK3, LAMTOR2, LAT, LCK, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MCM4, MEFV, MOGS, MSN, MTHFD1, MVK, MYD88, MYO5B, MYSM1, NBN, NCF1, NCF2, NCF4, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP12, NLRP3, NOD2, NSMCE3, ORAI1, OTULIN, PARN, PEPD, PGM3, PIK3CD, PIK3R1, PLCG2, PNP, POLA1, POMP, PRF1, PRKCD, PRKDC, PSMB8, PSTPIP1, PTPRC, RAB27A, RAG1, RAG2, RASGRP1, RBCK1, RFX5, RFXANK, RFXAP, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RORC, RPSA, RTEL1, SAMHD1, SBDS, SERPING1, SGPL1, SH2D1A, SKIV2L, SLC29A3, SLC35C1, SLC37A4, SLC46A1, SMARCAL1, SP110, SPINK5, SPPL2A, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TBK1, TCF3, TCN2, TICAM1, TLR3, TMC6, TMC8, TNFAIP3, TNFRSF1A, TPP2, TRAC, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNC93B1, UNG, USB1, VPS13B, VPS45, WAS, WIPF1, XIAP, ZAP70, ZBTB24, ADAM17, ALPI, AP1S3, AP3D1, BCL10, BCL11B, BLOC1S6, CD247, CD4, CD81, CD8A, CDC42, CFB, CFHR1, CFHR3, CFHR4, CFHR5, CFTR, CIB1, COL7A1, CR2, CTC1, DBR1, DEF6, DNASE1L3, EFL1, ERBIN, FCGR3A, FCHO1, FNIP1, FPR1, GIMAP5, GUCY2C, HAVCR2, IFNAR1, IGKC, IL17F, IL21, IL2RB, IL6R, IL6ST, IPO8, IRF3, IRF7, IRF9, ITPKB, IVNS1ABP, KRAS, LIG1, MBL2, MPEG1, MPO, NCKAP1L, NCSTN, NFE2L2, NLRP1, NOP10, NPC1, NRAS, OAS1, PAX1, PIK3CG, PLG, PMS2, POLD1, POLE, POLR3A, POLR3C, PRIM1, PSENEN, PSMA3, PSMB10, PSMB4, PSMB9, PTEN, PTPN2, RAC2, RC3H1, RECQL4, RELA, RHOH, RNF31, RNU7-1, SAMD9, SAMD9L, SASH3, SEC61A1, SLC39A7, SLC7A7, SLC9A3, SMARCD2, SNORA31, SOCS1, SPI1, SRP54, STXBP3, SYK, TAPBP, TBX1, TERC, TERT, TET2, TFRC, TGFB1, TINF2, TLR7, TNFRSF11A, TNFRSF13C, TNFRSF9, TOP2B, TRAF3IP2, TRIM22, USP18, WDR1, ZNF341, ZNFX1, AB13, ACTB, APOL1, ARHGAP42, ARHGEF1, ATG4A, BRCA1, BRCA2, C8G, CD28, CFHR2, CLCN7, CNBP, COLEC11, CSF2, CTNNBL1, CXCR2, ELF4, EPCAM, ERCC2, ERCC3, ERCC4, FAAP24, FANCF, FANCI, FANCM, FBF1, FBRS, FCGR1A, FCGR2A, FCGR2B, FCGR3B, FCGRT, FCN3, FERMT1, FOXM1, FPR2, FPR3, GAD1, GIMAP6, GTF2H5, HMOX1, HS3ST6, HYOU1, ICOSLG, IFNAR2, IFNG, IGHG2,

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IKZF3, IL12RB2, IL17A, IL18, IL18BP, IL22, IL23A, IL23R, IL31RA, IL37, IL6, IRAK1, IRF2BP2, IRF4, ITGAM, JAK1, KDM6A, KMT2A, KMT2D, LCP2, LRRC32, LRRC8A, LSM11, LYZ, MAP1LC3B2, MAPK8, MASP1, MASP2, MCM10, MED13L, MICA, MPI, MR1, MRE11, MS4A1, MSH6, MTPAP, MYOF, NBAS, NFAT5, NFKBID, NOS2, ODC1, OSTM1, PARP1, PDCD1, PLEKHM1, POLD2, POLE2, POLR3F, POU2AF1, PSEN1, PSMG2, PTPN22, RAD50, RANBP2, REL, RELB, RELN, RET, RGS10, RHOG, RNU4ATAC, SAMD3, SART3, SEMA3E, SH3BP2, SH3KBP1, SLC13A4, SNX10, STAT4, STAT5A, STN1, TBX21, TCIRG1, TGFBR1, TGFBR2, THBD, TIRAP, TLR8, TNFRSF13B, TNFRSF4, TNFSF11, TNFSF12, TNFSF13, TNIP1, TOM1, TRAF3, TSPAN14, TUBGCP3, UBA1, UNC119, WRAP53, ZC3HC1, ZFP36, ZNF34.

Variant classification as per ACMG guidelines:

Variant	A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).
Benign	A variant which is very likely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of Pathogenicity.
Likely Benign	A variant which is known not to be responsible for disease has been detected. Generally no further action is warranted on such variants when detected.
Pathogenic	A disease causing variation in a gene which can explain the patient's symptoms has been detected. This usually means that a suspected disorder for which testing had been requested has been confirmed.
Likely Pathogenic	A variant which is very likely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity.
Variant of Uncertain Significance	A variant has been detected, but it is difficult to classify it as either pathogenic (disease causing) or benign (non-disease causing) based on current available scientific evidence. Further testing of the patient or family members as recommended by your clinician may be needed. It is probable that their significance can be assessed only with time, subject to availability of scientific evidence.

Recommendations:

- Genetic counselling is advised for interpretation on the consequences of the variant(s).
- We recommend confirming the presence of these variants by Sanger Sequencing.

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- Segregation analysis of the variant by Sanger sequencing is recommended in affected member and unaffected member of the family.
- The results should be interpreted in the context of the patient's medical evaluation, family history and racial/ethnic background. Please note that variant classification and/or interpretation may change over time if more information available. Re-interpretation of multi gene next generation sequencing data is recommended on an annual basis and may be requested by a medical provider.
- For questions about this report, or for assistance in locating nearby genetic counselling services, please contact the Laboratory: contact@sngenelab.com.
- If results obtained do not match the clinical findings, additional testing should be considered as per referring clinician's recommendation.

Test Methodology:

- Genomic DNA isolated from Peripheral Blood, Saliva, Amniotic fluid, CVS, Cord Blood or any other standard source is used for NGS Library preparation.
- The libraries were sequenced to mean >85-100X coverage on Illumina sequencing platform. We follow the GATK best practices framework for identification of variants in the sample. The sequences obtained are aligned to human reference genome(Hg19).
- Clinically relevant mutations were annotated using published variants in literature and a set of diseases databases. Common variants are filtered based on allele frequency in 1000Genome, ExAC, gnomAD, dbSNP.
- Non-synonymous variants effect is calculated using multiple algorithms such as PolyPhen-2, SIFT, MutationTaster.
- Only non-synonymous and splice site variants found in the exome panel were used for clinical interpretation.
- Silent variations that do not result in any change in amino acid in the coding region are not reported.

Limitations:

- It should be noted that this test does not sequence all bases in a human genome, not all variants have been identified or interpreted, and this report is limited only to variants with evidence for causing or contributing to disease/clinical details provided to SN Gene Lab Pvt Ltd.
- Certain genes may not be covered completely and few mutations could be missed. Variants not detected by the assay may impact the phenotype.
- Triplet repeat expansions, translocations, large deletion or duplications and large copy number events are currently not reliably detected by next generation sequencing.

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- This assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, other gene rearrangements like inversion or translocation and does not detect single or multi-exon deletions or duplications.

References:

1. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology, Genetics in Medicine, 2015 May;17(5):405-24

2. Kalia S.S. et al., Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update(ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. Genet Med., 19(2):249-255, 2017.

3. Meyer, L.R., et al., The UCSC Genome Browser database: extensions and updates 2013. Nucleic Acids Res, 2013. 41(D1): p.D64-9.

4. McKenna, A., et al., The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data. Genome Res, 2010. 20(9): p. 1297-303

5. 1000 Genomes Project Consortium et al., A global reference for human genetic variation. Nature, 526(7571): 68-74, 2015.

6. Lek M. et al., Analysis of protein-coding genetic variation in 60,706 humans. Nature, 536(7616):285-91, 2016

7. McLaren, W., et al., Deriving the consequences of genomic variants with the Ensembl API and SNP Effect Predictor. Bioinformatics, 2010. 26(16): p. 2069-70.

---------- End Of Report ----------

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