
The miracle foundation

Advanced Pediatrics Centre
Postgraduate Institute of Medical Education and Research, Chandigarh
Report for Custom NGS Sequencing Assay

Name	Avleen Kaur	**Lab ID**	2942
Age/Sex	10 Yr/F	**CR No.**	202403967503
Sample	Blood	**Req. Date**	10-05-2025
Ward/OPD	Pediatric Medicine-Pediatric Hematology clinic (PHC)		

Clinical Details The patient, a suspected case of inherited bone marrow failure syndrome, was worked up for the targeted next generation sequencing.The said test comprises of a total of 33 genes. Analysis for variation was done by IonReporter tools.

Variant Details

Classification	Vus	**Locus**	chrX:48794094	**Reference**	C
Observed	T	**Type**	SNV	**Gene**	GATA1
Location	GATA1:exonic:NM_002049.3	**Length**	1	**Allele Frequency**	51.0%

Reported Variants

Gene.	GATA1	**Exon**	6	**Chromosol**	chrX:48794094
Classification.	Vus				

Comment

On data analysis, a heterozygous variant is seen in a female child in GATA-1 gene on X-chromosome. The variant is VUS as it fulfils PM2 and PP3 criteria and lacks evidence for further pathogenicity. Though the heterozygous variant in a female on an X-linked gene makes the patient a carrier, such individuals can present with variable degrees of mild to moderate thrombocytopenia or anemia due to skewed or random inactivation of X-chromosome. It is first advised to confirm this variant by sanger sequencing in index case and then screen any other siblings for a CBC and current variant to establish further pathogenicity.

Result Entry Date 08-05-2025 **Result Entered by** Deep Shikha(Jr.Lab.Technician)
Validation Date 09-05-25 11:20 AM **Validated by** Prateek Bhatia(Associate Professor)

(2942) U/C

Avleen Kaur - IBMFS_c9254_2025-05-30-10-14-27-684 Report

Advanced Paediatrics Centre

Postgraduate Institute of Medical Education and Research
Sector 12, Chandigarh
160012, India

Background

The patient, a suspected case of inherited bone marrow failure syndrome, was worked up for the targeted next generation sequencing. The said test comprises of a total of 33 genes. Analysis for variation was done by IonReporter tools.

Analysis

Avleen Kaur - IBMFS_c9254_2025-05-30-10-14-27-684

Ion Reporter Version 5 10	**Launched by** Dr Prateek Bhatia	**Analyzed by** Dr Prateek Bhatia	**Launched on** May-29-2025 09:31 PM
Workflow IBMFS_BAM r 0	**Annotations** All_GRCh38 r 0	**Reference** GRCh38, IAD151910_197_Designed	

Samples

Avleen Kaur - IBMFS

Gender Unknown	**Relationship** Proband

Reported Variants

Classification Vus	**Locus** chrX:48794094	**Genotype** C/T	**Filter** PASS
Ref C	**Observed Allele** T	**Type** SNV	**Genes** GATA1
Location GATA1 exonic NM_002049.3	**Length** 1	**% Frequency** 51.06	

Variant Details

Gene: GATA1 -- Exon: 6 --- chrX:48794094 --- NM_002049.3 --- Classification: Vus

Sample	Genotype	Amino Acid
Avleen Kaur - IBMFS	c.1172C>T	p.Thr391Met

* protein change takes into account changes at multiple genomic loci in same codon

Comments

On data analysis, a heterozygous variant is seen in a female child in GATA-1 gene on X-chromosome. The variant is VUS as it fulfils PM2 and PP3 criteria and lacks evidence for further pathogenicity. Though the heterozygous variant in a female on an X-linked gene makes the patient a carrier, such individuals can present with variable degrees of mild to moderate thrombocytopenia or anemia due to skewed or random inactivation of X-chromosome. It is first advised to confirm this variant by sanger sequencing in index case and then screen any other siblings for a CBC and current variant to establish further pathogenicity.

Sign-Off

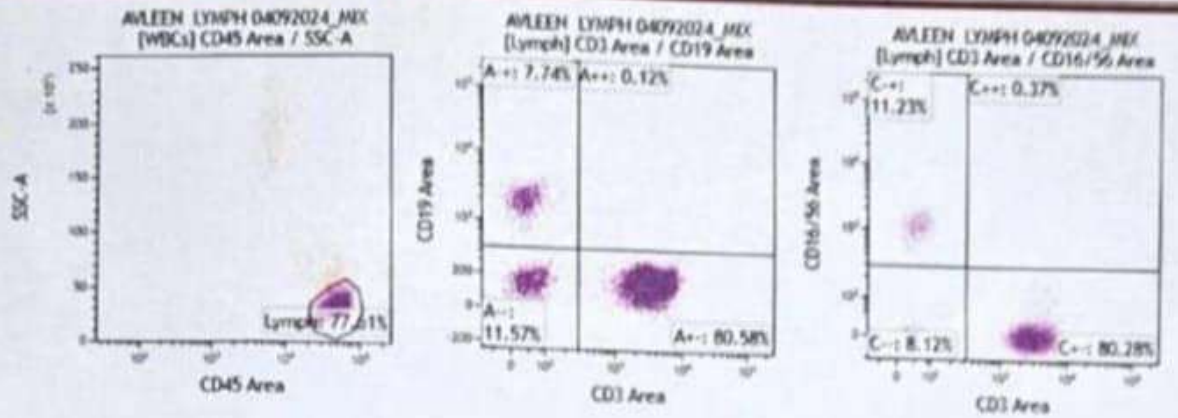
Pediatric Allergy and Immunology Laboratory
Advanced Pediatrics Centre
Dept of Pediatrics

50 Glorious years of PGIME

Postgraduate Institute of Medical Education and Research, Chandigarh

Name of the patient: Avleen Kaur **Age/Sex:** 10y/F **CR No:** 2025 0396 7503
Date: 04/09/2025 **Clinic:** APC **Clinical diagnosis:** Bi cytopenia

LYMPHOCYTE SUBSET



Total leucocyte count = 2230/cumm, Lymphocytes on DLC: LY:71%
Lymphocytes gated on SSc vs CD45 =77.61 %

Lymphocyte subset	Percentage	Absolute counts	Normal range
Lymphocyte count		1582	1900-3700
CD3+ T lymphocytes	80.58	1276	60-76 % (1200-2600)
CD19+ B lymphocytes	7.74	123	13-27 % (270-860)
CD56+dim+ NK cells	11.23	178	04-17 % (100-480)

Impression: Normal absolute counts of T and NK cells. Reduced absolute counts of B cells.

Advice:

Ig profile

Correlate clinically

Test performed: Mr.Satish/Ms.Gurjit Lab SR Dr. Saniya/Dr. Manpreet/Prof. Amit Rawat

PC
Lab No:- 2589/24

Postgraduate Institute of Medical Education and Research, Chandigarh

Department of Immunopathology

Research Block A, 4th Floor, Room No. 23

Indirect immunofluorescence antinuclear antibody test

Name	Avleen Kaur	Lab ID	55123/25
Age/Sex	10 Yr/F	CR No.	202503967503
Sample	Clotted Blood	Req. Date	17-04-2025
Ward/OPD	General Pediatric Ii And Hematology 5b		

Antinuclear antibody result

Interpretation

Antinuclear antibody Result	Negative
Antinuclear antibody Type	-
Antinuclear antibody Pattern	-
Antinuclear antibody Intensity	-

Test characteristics

Method	Indirect Immunoflourescence
Substrate	Hep-2 cells
Dilution	-

*ANA test has been interpreted as per recommendation/guidelines by the International Consensus on ANA patterns (ICAP), 2018

For details related to ANA patterns (now preferably called as anticellular antibody patterns) please refer to ICAP website: www.ANApatterns.org

Antigen likely to be associated

-

Disease likely to be associate

-

RB

Remarks

-

Advice

-

Entered by	Anita Meena(Junior Lab Technician)	Entered date	20-04-2025
Validated by	Yashwant Kumar(Professor)	Validated on	20-04-25 12:39 PM

Department of Hematology
Postgraduate Institute of Medical Education & Research, Chandigarh
REPORT OF BONE MARROW ASPIRATION / TREPHINE BIOPSY

Name: Avleen Kaur **Age / Sex:** 10 Yr/F **CR No:** 202503907503 **B.M.No:** P-559/24
Cl.I /c: **Ward:** Pediatric Medicine-E M G P M D **Dated:** 20-04-2025

Clinical Diagnosis:- Fever with skin ecchymosis with TCP and leukopenia. No history of lymphadenopathy and splenomegaly. No history of transfusion.

HEMOGRAM DETAILS **Hemogram No:** H-150

HB:- 9.8 gm/dl **Retic:** 1 % **PLT:** 6 X10^9/L **TLC:** 4.24 X10^9/L

DLC P- 9 L- 87 M- 3 E- 1 Ba- - Bl- - Pm-- My-- Mm-- nRBC- -

PBF: Normocytic normochromic red cells. Platelets are markedly reduced.

BONE MARROW FINDINGS

Particles: Particulate **NE:E Ratio** - **M:E Ratio** 1:1
Cellularity: Hypocellular

Blasts:	1	**Erythropoiesis:**	
Promyelocytes:	-	Normoblastic	
Myelocytes:	10	to megaloblastic. No ring sideroblasts noted.	
Metamyelocytes:	5	**Thrombopoiesis:**	
Polymorphs:	8	Decreased	
Lymphocytes:	24	-	
Monocytes;	-	**Others:**	
Eosinophils:	1	Erythroid precursors- 50% and Eo-Baso- 1%	
Basophils:		No hemophagocytosis noted.	
Plasma Cells:			

CYTOCHEMISTRY

LAP - **MPO** - **PAS** - **PERLS** 3+

TREPHINE BIOPSY REPORT **Trephine Biopsy No :**PTx-305/24

Bilateral trephine biopsies measuring 2.6- 1.8 cm shows markedly hypocellular marrow spaces with an overall cellularity of 5-10%. There is reduction of all the three hematopoeitic elements. No granulomas or immature collections seen.

Reticulin - -
Interpretation Hypocellular bone marrow
Advice 1. Kindly collect FISH cytogenetic report which will be available online

JR Dr Pavneet **SR** Dr Elgiva **Faculty** Dr Pulkit Rastogi
Validated On 02-05-25 11:53 AM **Validated By** Pulkit Rastogi(Assistant Professor)

Department of Hematology

Postgraduate Institute of Medical Education & Research, Chandigarh

REPORT OF BONE MARROW ASPIRATION / TREPHINE BIOPSY

Name: Avleen Kaur **Age / Sex:** 10 Yr/F **CR No:** 202403967503 **B.M.No:** P-559/24

Cl.I /c: **Ward:** Pediatric Medicine-E M G P M D **Dated:** 31/04/2025

Clinical Diagnosis:- Fever with skin ecchymosis with TCP and leukopenia. No history of lymphadenopathy and splenomegaly. No history of transfusion.

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DLC P- 9 **L-** 87 **M-** 3 **E-** 1 **Ba-** - **Bl-** - **Pm-** - **My-** - **Mm-** - **nRBC-** -

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Myelocytes: 10

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Lymphocytes: 24

Monocytes; -

Eosinophils: -

Basophils: -

Plasma Cells: -

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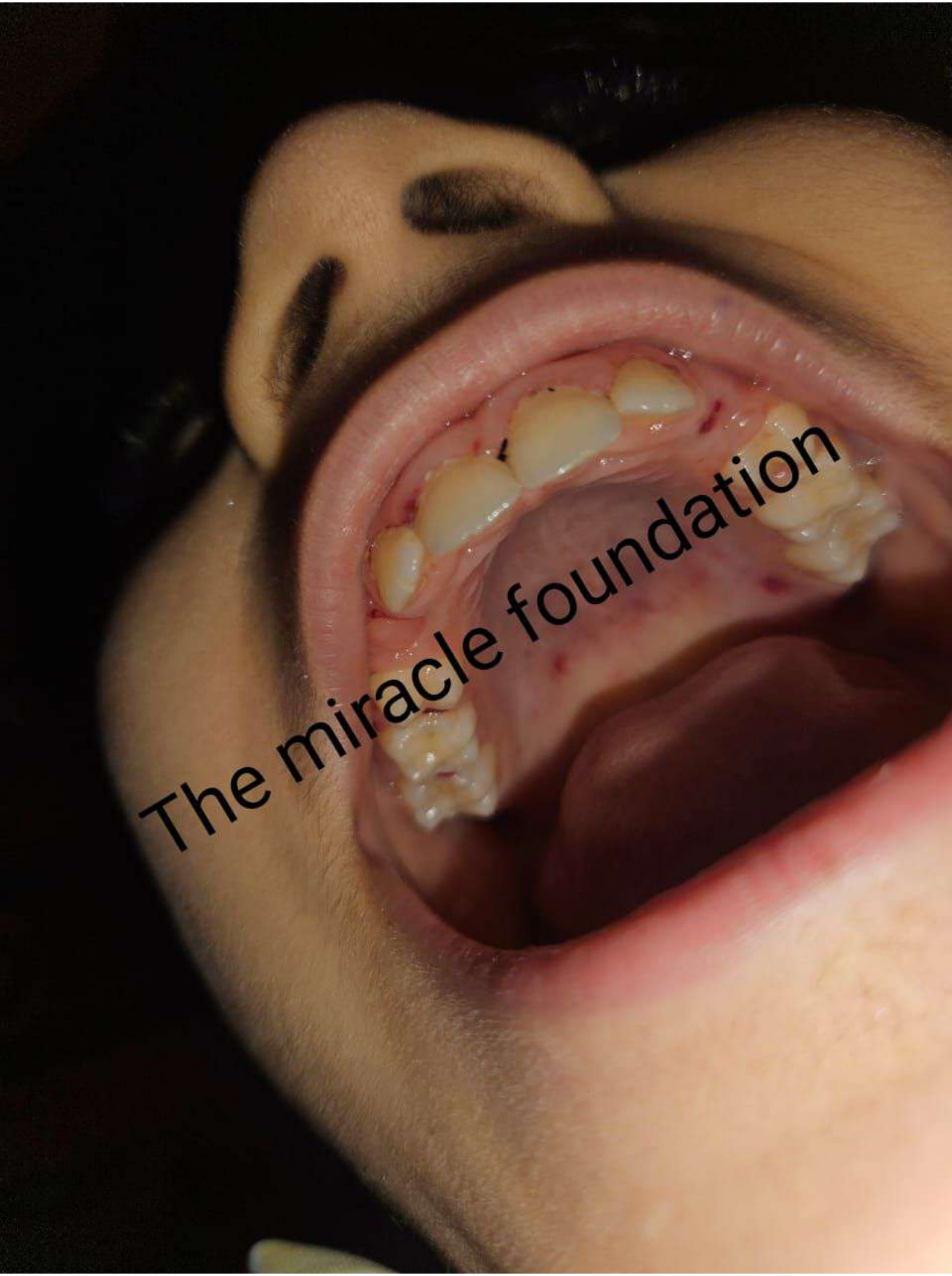
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भारत सरकार

Government of India

आधार

Avleen Kaur

Date of Birth/DOB: 12/08/2014

Female/ FEMALE

Download Date: 21/12/2020

Issue Date: 28/11/2020

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7075 0934 1619

VID : 9169 8351 7677 9474

मेरा आधार, मेरी पहचान