Histiocytosis-lymphadenopathy plus syndrome on follow-up of a child with secondary hemophagocytic lymphohistiocytosis.

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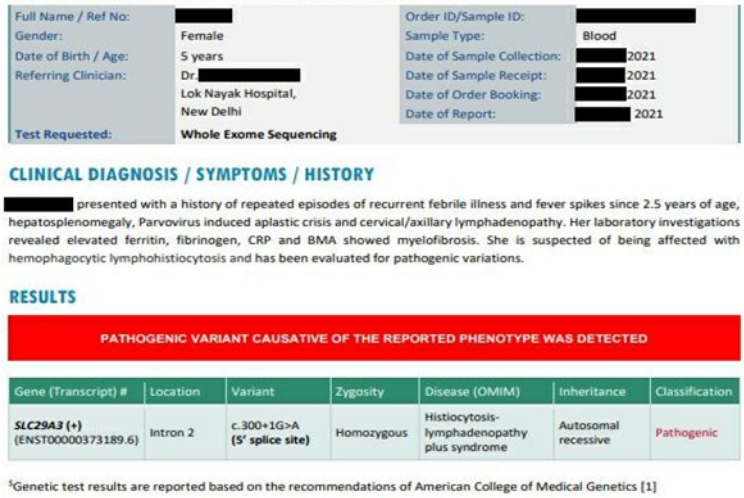

Full Name / Ref No:	[redacted]	Order ID/Sample ID:	[redacted]
Gender:	Female	Sample Type:	Blood
Date of Birth / Age:	5 years	Date of Sample Collection:	[redacted]2021
Referring Clinician:	Dr. [redacted]	Date of Sample Receipt:	[redacted]2021
	Lok Nayak Hospital, New Delhi	Date of Order Booking:	[redacted]2021
		Date of Report:	[redacted] 2021
Test Requested:	**Whole Exome Sequencing**		

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

[redacted] presented with a history of repeated episodes of recurrent febrile illness and fever spikes since 2.5 years of age, hepatosplenomegaly, Parvovirus induced aplastic crisis and cervical/axillary lymphadenopathy. Her laboratory investigations revealed elevated ferritin, fibrinogen, CRP and BMA showed myelofibrosis. She is suspected of being affected with hemophagocytic lymphohistiocytosis and has been evaluated for pathogenic variations.

RESULTS

PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

Gene (Transcript) #	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
SLC29A3 **(+)** (ENST00000373189.6)	Intron 2	c.300+1G>A **(5' splice site)**	Homozygous	Histiocytosis-lymphadenopathy plus syndrome	Autosomal recessive	Pathogenic

[$]Genetic test results are reported based on the recommendations of American College of Medical Genetics [1]