For Female

Name	: **Baby DUMMY-4**			**Collected**	: **8/9/2016 6:50:00PM**
				Received	: **8/9/2016 7:06:37PM**
Lab No.	: **106120076**	**Age: 5 Years**	**Gender: Female**	**Reported**	: **27/10/2016 12:40:44PM**
A/c Status	: **P**	**Ref By : SELF**		**Report Status**	: **Final**

Test Name	Results	Units	Bio. Ref. Interval
NIEMANN PICK DISEASE, QUANTITATIVE, BLOOD (Enzyme Assay)			
Patient Value	**1.00**	nmol/hr/mg	>3.00
Control Value	6	nmol/hr/mg	

Impression:-Deficient Sphingomyelinase enzyme activity.

Interpretation

SPHINGOMYELINASE ACTIVITY	REMARKS
>3	Normal activity
1.5-3	Possibility of carrier state likely
<1.5	Deficient activity

Note

1. Results should be clinically correlated as individual / biological variations can affect the test results
2. Genetic counseling available with prior appointment at Department of Genetics, National Reference Lab, New Delhi

Comments

Niemann Pick disease (Types A & B) is a lysosomal storage disease caused by deficiency of enzyme Sphingomyelinase. It is inherited as an autosomal recessive disorder.

Type A disease is characterized by jaundice, progressive loss of motor skills, feeding and learning difficulties and hepatosplenomegaly. It usually presents within 1-4 months of age and death occurs by 3 years.

Type B disease is milder though variable in clinical presentation. Most of these patients do not have neurological involvement and survive upto adulthood.

Type C disease is a lysosomal lipid storage disease presenting usually in middle to late childhood. It is characterized by vertical gaze palsy, ataxia, dystonia, behavioural problems and dementia.

Dr. Manjeet Kaur
PhD
HOD Genetics

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