SAVINGS ARE NOT ALWAYS SAFE

DEPARTMENT OF INSTITUTE OF CHILD HEALTH

- Dr S Elilarasi.
- Dr Kalpana.S
- Dr B Sarath Balaji.

- 5yr old male child
- 3rd born of second degree consanguinous marriage
- Referred as a case of interstitial lung disease
- c/o
  - fever
  - cough
  - breathlessness

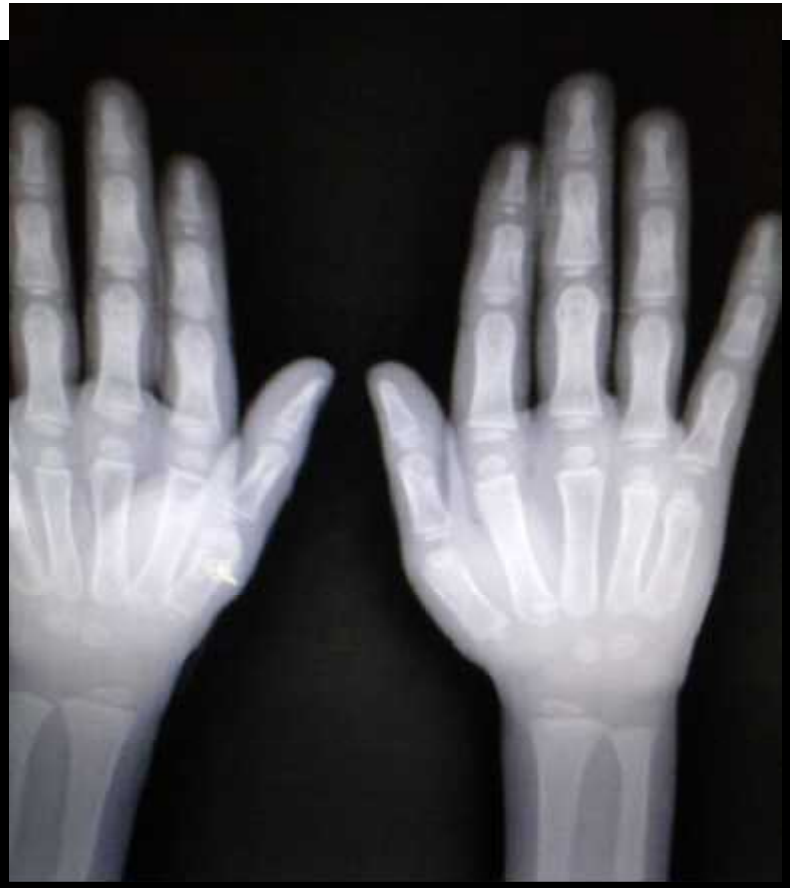
3 weeks

C

- Conscious and oriented
- Developmentally normal
- no regression
- Pallor
- Frontal bossing
- Depressed nasal bridge
- No clubbing
- Not icteric
- Skin- normal

- **Height - 92 cm**
- **< 3rd centile**

- **Weight – 12 kg**
- **< 3rd centile**



BONE AGE – 2 yrs

- CVS: normal

- RS: RR-30/min
  b/l crepitations +
  Sao2 – 97%
- Abdomen : distended,
  Liver span 8cm,
  not tender,firm
  Spleen 7 cm , firm

- CNS: clinically normal

- no significant illness in the past
- 2 elder siblings-normal

BLOOD CO

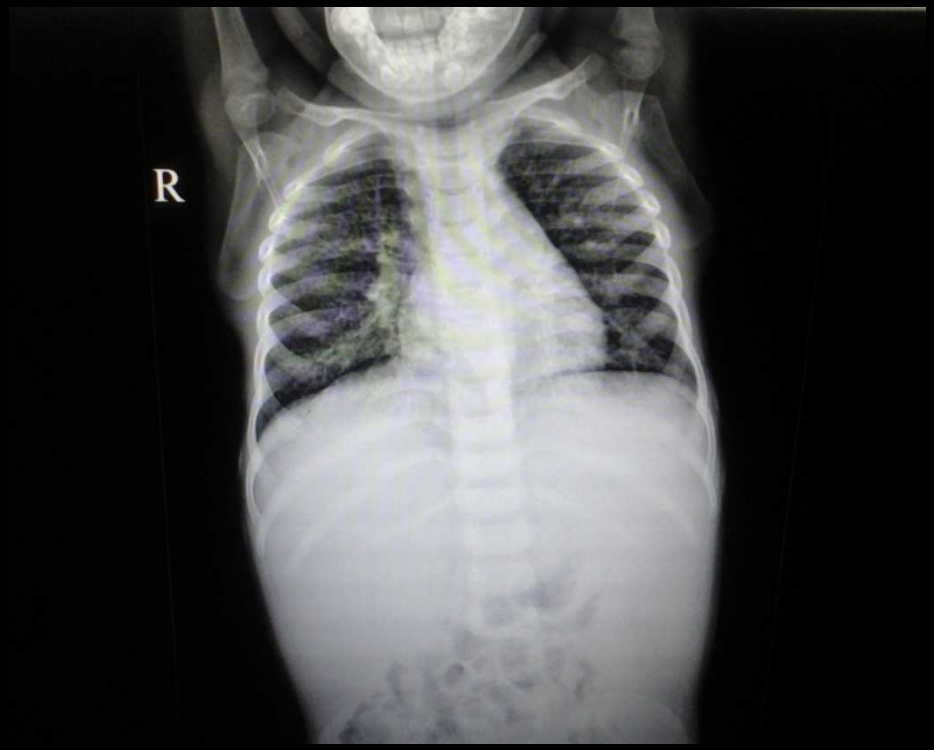
12/5/15	13/5/15	22/5/15
3,400	4,900	8,400
P26 L74	P41 L59	P40 L53 E07
10.8	8.4	11.5
30.5		34.7
1.23 lakhs	1.8 lakhs	2.26 lakhs.
< 9 / 27 mm		< 6 mm / 18 mm

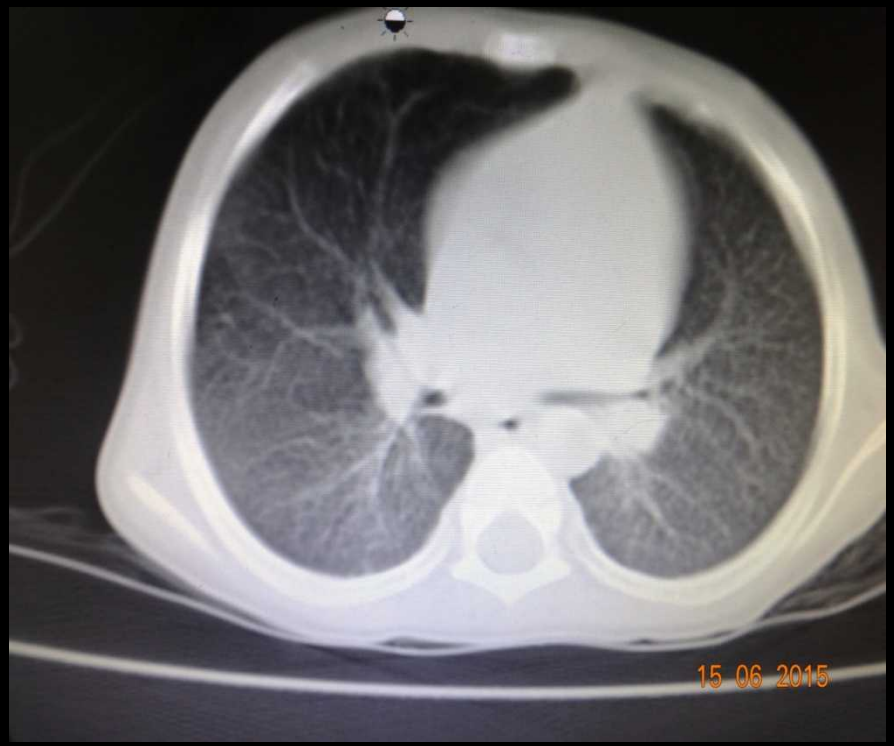
mic, moderate anisocytosis / 13/5/15 — repeat

INVESTIGA

- RFT,LFT – normal
- P.smear- moderate hypochromic mild anisocytic, and atypical lymphocytes,
- USG abdomen : hepatosplenomegaly ,no free fluid
- Mantoux and RGJ negative

CLI





PROBLEMS

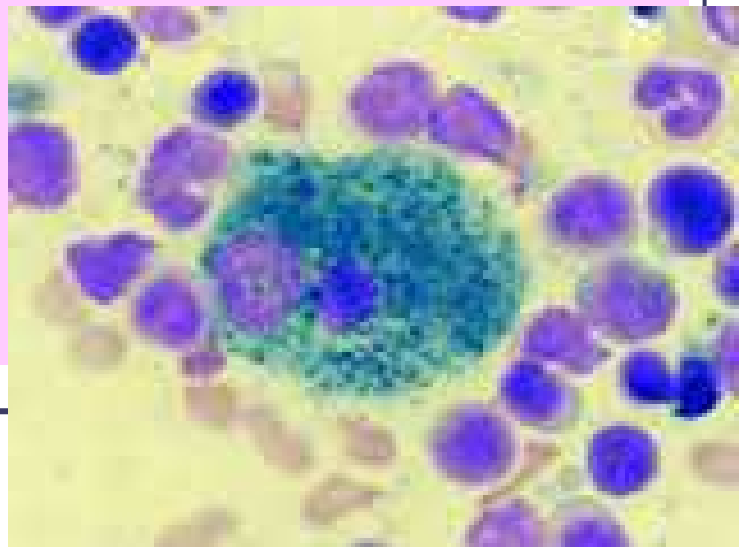
- SHORT STATURE
- HEPATOSPLENOMEGALY
- ILD
- FRONTAL BOSSING
- PANCYTOPENIA

DIFFERENTI

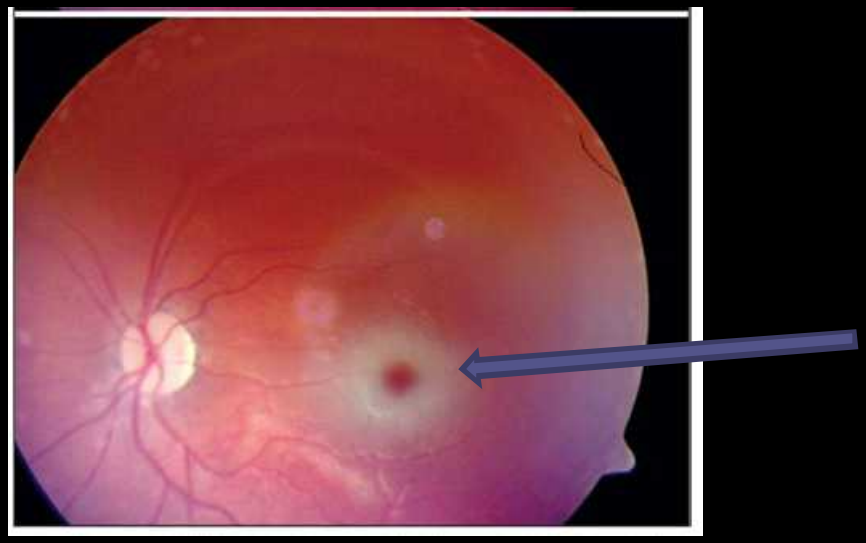
- Hemolytic anaemia — HB ELECTROPHORESIS
- Wilson's disease — SERUM CERULOPLASMIN, 24 HR URINARY COPPER
- LCH — X RAY SKULL ,POST AURICULAR BIOPSY
- STORAGE DISORDER

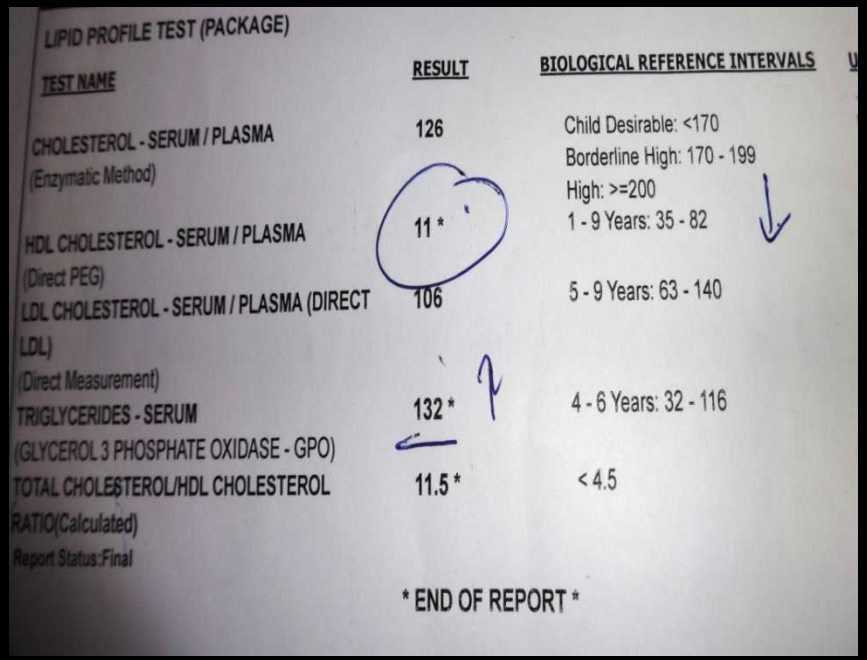
BONE MARROW ASPIRATION

- LARGE FOAMY MACROPHAGES
- SEA BLUE HISTIOCYTES



FUNDUS





LIPID PROFILE TEST (PACKAGE)

TEST NAME	RESULT	BIOLOGICAL REFERENCE INTERVALS
CHOLESTEROL - SERUM / PLASMA (Enzymatic Method)	126	Child Desirable: <170 Borderline High: 170 - 199 High: >=200
HDL CHOLESTEROL - SERUM / PLASMA (Direct PEG)	11 *	1 - 9 Years: 35 - 82
LDL CHOLESTEROL - SERUM / PLASMA (DIRECT LDL) (Direct Measurement)	106	5 - 9 Years: 63 - 140
TRIGLYCERIDES - SERUM (GLYCEROL 3 PHOSPHATE OXIDASE - GPO)	132 *	4 - 6 Years: 32 - 116
TOTAL CHOLESTEROL/HDL CHOLESTEROL RATIO(Calculated)	11.5 *	< 4.5

Report Status:Final

* END OF REPORT *

- HDL is decreased
- Elevation of LDL and triglycerides

NIEMANN PICK DISEASE TYPE B

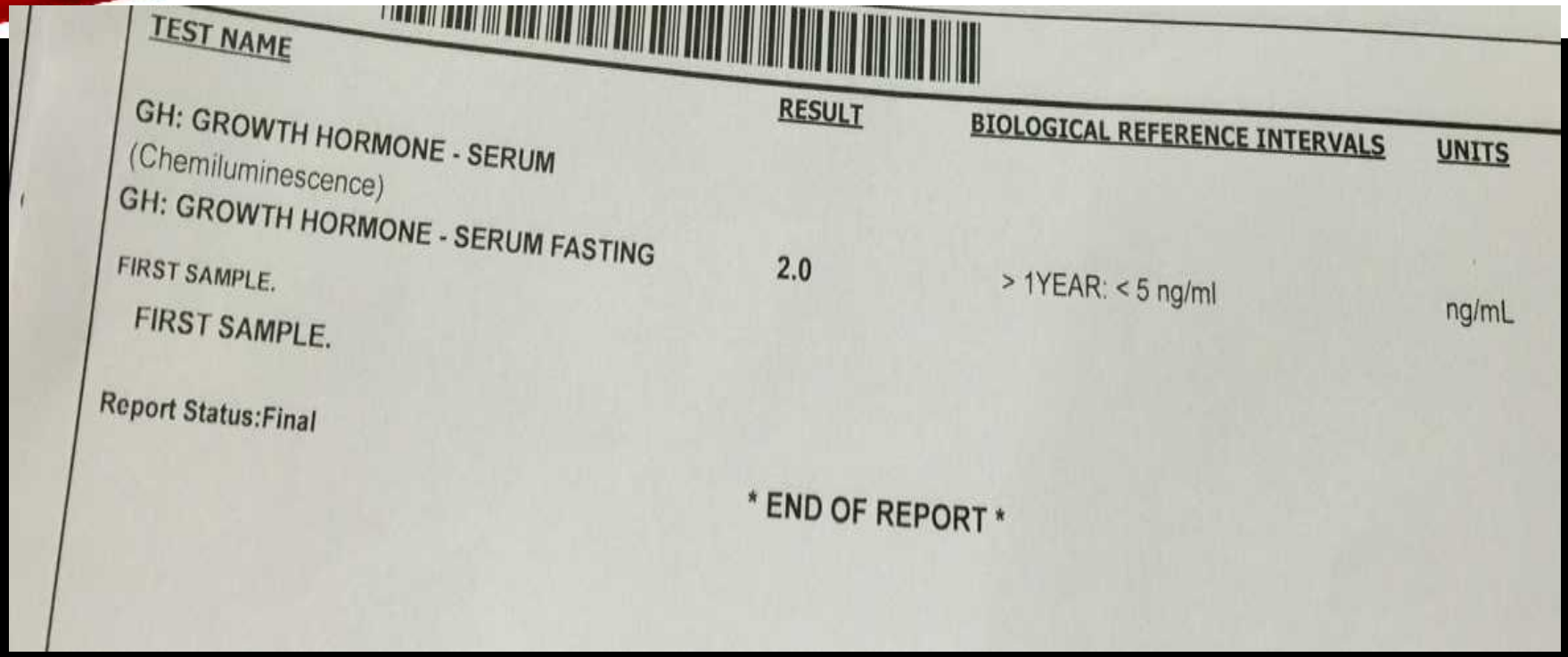
ENZYME ST

Lysosomal Enzymes Study From Leucocytes

ENZYMES	Result	Normal Range
Glycolipids and lipids		
Sphingomyelinase **(Niemann Pick Disease A & B)**	: 0.69	1.8 - 6.9 nmol/17hr/mg protein
β-galactosidase **(GM1 Gangliosidosis)**	: 68.4	32.5 - 206.5 nmol/hr/mg protein
Plasma Chitotriosidase	: 267.1	28.66 - 62.94 nmol/ml/hr **7275.59 ± 6161.72 (NPD A/B)** **28239.2 ± 20186.7 (Gaucher Disease)**

Remarks : Lysosomal enzyme study was carried out from leucoytes using 4-MU specific substrate for Sphingomyelinase. β-galactosidase was used as a reference enzyme. Master Sanjay has shown reduced activity of Sphingomyelinase (20% of the mean enzyme activity) with normal activity of β-galactosidase. **This suggests that Sanjay is highly likely to be affected with NPD-A/B disease.** This is an autosomal recessive genetic disorder with recurrent risk of 25% in each pregnancy. Family is advised for genetic counseling and prenatal diagnosis at 11-16 wks of next pregnancy.

GP



TEST NAME	RESULT	BIOLOGICAL REFERENCE INTERVALS	UNITS
GH: GROWTH HORMONE - SERUM (Chemiluminescence)			
GH: GROWTH HORMONE - SERUM FASTING	2.0	> 1YEAR: < 5 ng/ml	ng/mL
FIRST SAMPLE.			
FIRST SAMPLE.			

Report Status:Final

* END OF REPORT *

- FASTING AND FOLLOWING CLONIDINE STIMULATION

TEST NAME	RESULT	BIOLOGICAL REFE
GH: GROWTH HORMONE - SERUM		
(Chemiluminescence)		
Growth Hormone After Arkamine	1.0 *	3.6 - 39.9
THIRD SAMPLE.		
THIRD SAMPLE.		

Report Status:Final

*** END OF REPORT ***

NIEM

DISEASE TYPE B WITH GROWTH HORMONE DEFICIENCY

NIEMANN P

- Deficient activity of acid sphingomyelinase ,a lysosomal enzyme
- Pathologic accumulation of sphingomyelin in monocyte and macrophage system
- Autosomal recessive

TYPE B

- Late onset , non neuropathic
- Better prognosis
- Detected during childhood
- Survival adulthood typical
- Pulmonary symptoms common

COMPLIC

- **Splenic rupture- avoid contact sport**
- **hypersplenism**
- **Liver failure-transplantation**
- **Recurrent chest infections**
- **Progressive pulmonary disease- ygen dependance**

PULMONARY M
NIEMAN

- Alveolar infiltration- interstitial lung disease
- Decreased diffusion
- Progressive pulmonary disease- oxygen dependance
- Susceptible to recurrent infections and mycobacterial infections
- Bronchiectasis
- Cor pulmonale

Cases reported

- PUBMED
- **Evidence of pol**
  type B
- Strisciuglio P, Di Maio S, Parenti G, Franzese A, Lubrano P, Mariano A, Andria G.
- **Author information**
- **Abstract**
- We report a girl with Niemann-Pick disease type B in whom short stature was recorded over a long period. Association of short stature with the ***presence of a polyglandular involvement*** in this patient is discussed.

Landas et al. (1985) reported a 48-year-old woman with debilitating and eventually fatal coronary artery disease and hepatosplenomegaly in whom multiorgan infiltration by sea-blue histiocytes was the consequence of Niemann-Pick disease type B. Strisciuglio et al. (1987) found evidence of involvement of *multiple endocrine glands in a patient with type B Niemann-Pick disease and growth failure.*

STORAGE DISORDERS

- Enzyme replacement therapy
- Hurler
- Hunter
- Fabrys
- Gauchers
- Maroteux Lami
- Pompes

- Substrate reduction therapy
- miglustat
- Nitisinone

NEW IN NIEMANN PICK DISEASE

TYPE B

Bone marrow transplant

reduction of

liver and spleen,

storage cells, radiological

detected lung involvement

Enzyme replacement therapy

- **Phase 2 trials**
- **OLIPUDASE ALPHA**

- **TYPE A -Orthotopic liver transplant**
- **TYPE C- Substrate reduction therapy**


Thank You