BSH 2020 VIRTUAL
9 -14 NOVEMBER
British Society for Haematology
Listening • Learning • Leading
60 1960–2020

DIAGNOSIS OF NIEMANN-PICK DISEASE TYPE C ON BONE MARROW BIOPSY: A RARE EVENT

S JAHANGIR[1],S KAMRAN[1], S KHAN[1]
1 Shifa International Hospital,Islamabad, Pakistan

Shifa International Hospitals Ltd.

INTRODUCTION

- Niemann-Pick type C disease is a lysosomal storage disorder characterized by an inability of the body to transport cholesterol and fatty acids inside of cells.[1]
- Lysosomes are organelles that contain digestive enzymes involved in breakdown of proteins, nucleic acids, carbohydrates and lipids.
- Four basic types are identified categorized on basis of genetics and symptomatology.
- Niemann-Pick type C disease is due to mutation in NPC 1 gene located on long arm of chromosome 18(18q11.2) and NPC 2 gene located on long arm of chromosome 14 (14q24.3).[2]

AIM

- Diagnosis of Niemann Pick disease involves detailed history, thorough physical examination along with laboratory investigations such as blood based testing for biomarkers(oxysterols, lysosphingolipids, bile acid metabolites) and genetic analysis.
- Psychiatric manifestations often mislead physicians towards dementia and schizophrenia.
- Thorough workup should be carried out in case of a strong clinical suspicion.

CASE PRESENTATION

- A 39 year old patient presented with fever and abdominal pain .
- Abdominal examination revealed tenderness in right iliac fossa while ophthalmic exam confirmed vertical gaze palsy.
- Patient also reported behavioral changes and depressive episodes for the past two years.
- Complete blood count showed thrombocytopenia.

RESULTS

- Bone marrow aspirate showed a normocellular specimen with active granulopoiesis and erythropoiesis but highlighted few scattered large histiocyte-like cells having abundant cytoplasm with soap-bubble like appearance and morphology suggestive of storage cells.
- Bone marrow biopsy further confirmed the diagnosis by highlighting histiocyte - like cells .
- These cells were negative for periodic acid Schiff stain.
- On immunohistochemistry, these cells were strongly positive for CD68.

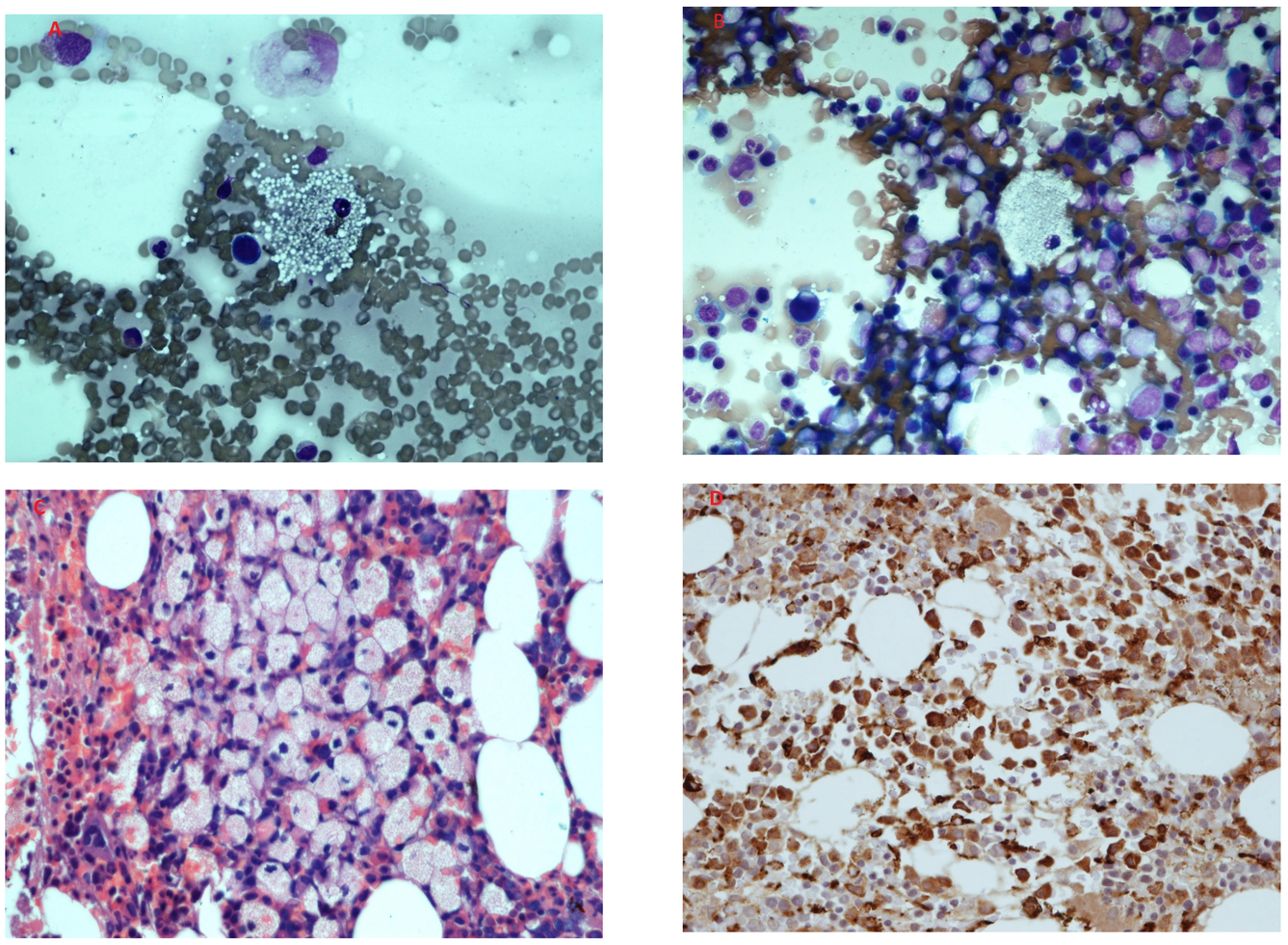

Figure A and B : Bone marrow aspirate(Leishman stain) showing diffuse infiltration by foam cells. These foam cells show abundant cytoplasm filled with numerous small vacuoles.
Figure C: Bone marrow trephine section shows marked increase in histiocyte-like cells
Figure D: Strong positivity seen in CD 68 immunostain.

CONCLUSIONS

Patients with progressive vertical supranuclear gaze palsy,ataxia, dystonia and dementia should be screened for underlying storage disorders as early diagnosis helps in management of these patients.

ACKNOWLEDGEMENT

Department of Pathology and Laboratory Medicine, Shifa International Hospital, Islamabad

REFERENCES

1)Lad M, Thomas RH, Anderson K, Griffiths TD, Niemann pick type C: comptemporary diagnosis and treatment of a classical disorder. Practical neurology. 2019 Oct1;19(5):420-3.
2)Evans WR,Hendriksz CJ. Niemann Pick type C disease- the tip of the iceberg? A review of neuropsychiatric presentation, diagnosis and treatment. BJPsych bulletin . 2017 Apr;41(2):109-14.

CONTACT INFORMATION

sehreenjahangir@yahoo.com
drshawana33@gmail.com
samarif1115@gmail.com