CASE REPORT

Gaucher Disease Involving Virchow’s Lymph Node: a Case Report

Dmitry A. Zinovkin1, Md Zahidul Islam Pranjol2, Dmitry Kravchenko3, Olga Kravchenko4, Vadim Kudryashov4

1 Department of Pathology, Gomel State Medical University, Gomel, Belarus
2 William Harvey Research Institute, Barts & The London School of Medicine & Dentistry, Queen Mary University of London, London, England
3 Republican Research Center for Radiation Medicine and Human Ecology, Gomel, Belarus
4 Regional Clinical Oncological Dispensary, Gomel, Belarus

Correspondence:
Dmitry A. Zinovkin, Department of Pathology, Gomel State Medical University, 531 Lange Str., 246000 Gomel, Belarus
E-mail: zinovkin2012@gmail.com
Tel: +375447413056

Received: 27 Feb 2018
Accepted: 22 May 2018
Published online: 06 Aug 2018
Published: 29 Nov 2018

Key words: Gaucher disease, Gaucher cells, N370S, CD68, Virchow’s lymph node


INTRODUCTION

Gaucher disease is one of the most common glycolipid storage disorders, caused by an inherited deficiency of the lysosomal enzyme β-glucocerebrosidase, leading to accumulation of the substrate glucocerebroside in the cells of the monocyte-macrophage system. Pancytopenia, hepatosplenomegaly and skeletal complications are hallmarks of Gaucher disease. This disease is traditionally classified into three broad phenotypic categories: type 1 (non-neuronopathic disease); type 2, fulminant neuronopathic disease that is fatal during infancy; and type 3, chronic neuronopathic disease, that usually results in death in childhood or early adult life.1

CASE REPORT

Gaucher disease is a metabolic storage disorder caused by a mutation in the lysosomal enzyme B-glucocerebrosidase. This disease is usually manifested in newborn infants, however, an exceptional case of this disease in an adult has been recently reported. A 21-year-old Caucasian patient was diagnosed with Gaucher disease, demonstrating Virchow’s lymphatic node enlargement and mild splenomegaly. A familial link to this disease was also found. Macrophage infiltration was observed in the affected Virchow’s lymph node which is not a classic sign of Gaucher disease. DNA analysis and a whole blood count also suggested a manifestation of this disease. In summary, this is the first study to report such a case of Gaucher disease in an adult female patient, which may suggest an asymptomatic characteristic of this condition and an importance of the presence of Gaucher cells in the enlarged Virchow’s lymph node.

A 21-year-old female Caucasian patient was admitted with symptoms of malaise, fatigue and pain in bones. Family history revealed that two relatives (father and sister) suffered from Gaucher disease. Physical examination demonstrated an enlarged Virchow’s lymphatic node and mild splenomegaly. Complete blood count results are as follows: RBC – 3.31×10⁶/µl, MCV – 86.1 fl, RDW – 17.2%, HCT – 28.5%, hemoglobin – 91.9 g/l, MCHC – 27.7 g/l, PLT – 320×10³/µl, thrombocrit – 0.29%, MPV – 9.3 fl, PDW – 17.2%; WBC – 9.41×10³/µl, granulocytes: band forms – 2%, neutrophils – 46%, eosinophils – 1%, basophils – 1%; lymphocytes – 47%; and monocytes – 3%. The assessment of Virchow’s node biopsy revealed areas of normal lymph node efface-
ment by large round cells (Fig. 1A) with lattice-like cytoplasm and eccentric nuclei, which characterize Gaucher cells (Fig. 1B). To differentiate between Gaucher disease, Hodgkin’s lymphoma and signet-cell carcinoma, an immunohistochemical study was performed using anti-pCK, anti-CD15, anti-CD30, anti-fascin, anti-Epstein-Barr virus and anti-CD68 antibodies. Only CD68 staining was positive in cells with foamy cytoplasm (Fig. 1C), suggesting a presence of macrophages.

DNA sequence analysis displayed mutation of glucocerebrosidase gene located in chromosome 1q21 and homozygous for the N370S mutation. Interestingly, PCR data did not present homozygous L444P mutations in the glucosylceramidase β gene, which is a marker of Gaucher disease. Glucocerebrosidase enzyme activity in the peripheral blood leukocytes was 29%, which is higher than the normal range: 0%-15%. Concluding all the data, the patient was diagnosed with type 1 Gaucher disease and was prescribed Cerezyme® as a therapy. Patient was discharged from hospital afterwards with significant clinical improvement.

DISCUSSION

Type 1 (non-neuronopathic) Gaucher disease accounts for more than 90% all Gaucher disease patients. Its prevalence worldwide is 1 in 50,000 to 100,000, but it is as high as ~1 in 850 in individuals of Ashkenazi heritage. The broadest phenotypic spectrum in Gaucher disease with respect to age of onset, rate of progression, and organs affected occurs in type 1 Gaucher disease. Many manifestations of Gaucher disease, some not uncommon and others very rare, cannot be explained by storage per se; examples include immunologic abnormalities, increased prevalence of certain malignancies with relative paucity of others, neurologic comorbidities (peripheral neuropathy and Parkinsonism), calcification of cardiac valves, and pulmonary hypertension.

Gaucher-like cells also can be found in some hematologic abnormalities, such as chronic lymphocytic leukemia and Hodgkin’s lymphoma. However, we did not observe specific histopathological

Figure 1. Biopsy of Virchow’s lymph node. A: Lymph node structure is completely effaced by a diffuse Gaucher cells infiltration of lymphoid tissue at the big area. Stain: hematoxylin and eosin. Magnification: ×40; B: Gaucher histiocytes with foamy cytoplasm and eccentric situated nuclei. Stain: hematoxylin and eosin. Magnification: ×200; C: CD68 strong expression by Gaucher cells infiltrating lymph node structures. Immunohistochemistry: anti-CD68 primary antibody. Magnification: ×200.
was not observed in the present case study, we assume that it could be a specific characterization for manifestation of Gaucher disease in adult patients. Nevertheless, in this case thrombocytopenia was not presented at diagnosis, which is a symptom of Gaucher disease. We agree with the suggestion that Gaucher disease presented by intraosseous conditions in adult age may be a prognostic factor of this disease.

CONCLUSION

To our knowledge, this is the first study to report an involvement of the Virchow’s node by Gaucher disease. Gaucher disease may remain asymptomatic for a long period of time and ultimately be diagnosed in adult age. Therefore, adult patients with foamy macrophages infiltration of lymph nodes and family history of Gaucher disease must be diagnosed with this disease condition and treated.

REFERENCES

Болезнь Гоше, поражающая железу Вирхова: клинический случай

Дмитрий А. Зиновкин1, Захидул Ислам Пранджол2, Дмитрий Кравченко3, Олга Кравченко4, Вадим Кудряшов4

1 Кафедра „Патология“, Республиканский исследовательский центр радиологии и экологии, Гомельский государственный медицинский университет, Гомель, Беларусь
2 Исследовательский институт „Уильям Харви“, Институт Бартс и Лондонская школа медицины и стоматологии, Лондонский университет королевы Марии, Лондон, Англия,
3 Республиканский исследовательский центр радиологии и экологии, Гомель, Беларусь
4 Региональный клинический онкологический диспансер, Гомель, Беларусь

Адрес для корреспонденции: Дмитрий А. Зиновкин, Кафедра „Патология“, Республиканский исследовательский центр радиологии и экологии, Гомельский государственный медицинский университет, ул. „Ланге“ № 531, Гомель, Беларусь E-mail: zinovkin2012@gmail.com Tel: +375447413056

Дата получения: 27 февраля 2018
Дата приемки: 22 мая 2018
Дата онлайн публикации: 06 августа 2018
Дата публикации: 20 ноября 2018

Ключевые слова: болезнь Гоше, клетки Гоше N370S, CD68, железа Вирхова

Болезнь Гоше является заболеванием метаболического накопления, вызванное мутацией в лизосомальном ферменте β-глюкоцереброзидазы. Эта болезнь обычно встречается у новорожденных, но сообщается о крайне редком случае заболевания взрослого человека. 21-летнему пациенту белой расы был поставлен диагноз болезни Гоше, проявляющейся увеличением железы Вирхова и лёгкой формой спленомегалии. Установлена также наследственная связь заболевания. Инфильтрация макрофагами наблюдалась в поражённой железе Вирхова, что не является классическим признаком болезни Гоше. Анализ ДНК и полный анализ крови тоже показали проявление данного заболевания. В целом, это первое исследование, в котором сообщается о таком случае болезни Гоше у взрослой женщины, что может говорить о бессимптомной особенности этого заболевания и о важности наличия клеток Гоше в увеличенной железе Вирхова.