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Type I Gaucher disease in adults with immune dysfunction: a case report and review of the literature

Quan Liu

liuquan123111@163.com

Huaibei People's Hospital (Huaibei People's Hospital Affiliated to Bengbu Medical University)

Li Wei

Huaibei People's Hospital (Huaibei People's Hospital Affiliated to Bengbu Medical University)

Ling Ge

Huaibei People's Hospital (Huaibei People's Hospital Affiliated to Bengbu Medical University)

Luying Zhao

Suixi County People's Hospital

Case Report

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Abstract

Background Gaucher disease (GD) is a rare autosomal recessive lysosomal storage disorder caused by *GBA1* gene mutations, and immune dysfunction is one of its important clinical manifestations, but the alterations of natural killer (NK) and natural killer T (NKT) cells in adult type I GD are rarely reported. This study aims to report a case of adult-onset type I GD with immune dysfunction, explore its unique immune phenotype, and provide evidence for clinical immune monitoring and individualized treatment of GD.

Case presentation A 58-year-old male of Han ethnicity was admitted due to incidentally detected splenomegaly and pancytopenia. Abdominal ultrasound revealed marked splenomegaly. Laboratory evaluation demonstrated pancytopenia with mild anemia. Bone marrow biopsy showed scattered histiocytic sheets, and bone marrow smears revealed numerous typical Gaucher cells (9.0% of nucleated cells). Glucocerebrosidase (GCase) activity was reduced to 5.92 nmol/h/mL. *GBA1* gene sequencing revealed a homozygous mutation in exon 7: NM_000157.4:c.907C > A (p.Leu303Ile), confirming the diagnosis of type I GD. Immunological assessment outcomes showed decreased serum levels of total protein, globulin, IgG, IgA, complement C3 and C4, as well as a marked reduction in NK cells (4.44%, 21 cells/μL) and mild elevation in NKT cells (11.64%, 55 cells/μL) in peripheral blood lymphocyte subsets.

Conclusions Type I GD is associated with diverse immune dysfunction, and this case presents a unique immunophenotype of NK cell depletion combined with mild NKT cell elevation, which enriches the immune phenotype spectrum of adult GD. Comprehensive immunological evaluation including lymphocyte subset analysis (especially NK and NKT cells) should be routinely performed in GD patients to guide individualized treatment and assess the risks of infection and malignancy.

Introduction

Gaucher disease (GD), also known as glucocerebrosidosis, is an autosomal recessive lysosomal storage disorder. Mutations in the gene encoding glucocerebrosidase (GCase) result in reduced or absent GCase activity, leading to excessive accumulation of its substrate, glucocerebroside, within monocytes/macrophages (Mo/Mφ). This causes the formation of characteristic “Gaucher cells,” primarily affecting the spleen, liver, bone marrow, and lymph nodes. The most prominent symptoms are hepatosplenomegaly, with splenomegaly being the most common. Some patients experience bone pain and growth retardation, and the disease may even compromise immune function [1]. Patients may also face increased risks of autoimmune dysfunction, monoclonal gammopathy, and multiple myeloma. GD causes abnormal accumulation of glucosylceramide (GL1) and its deacetylated form, lyso-GL1, leading to significantly elevated levels that mediate immune dysfunction [2]. In recent years, GD-related immune dysfunction has garnered increasing attention, particularly regarding peripheral blood lymphocyte subset profiling in GD patients.

However, reports on alterations in natural killer T (NKT) cells remain scarce. This study examines a case of adult type I GD with pronounced immune dysfunction admitted to our hospital, characterized by

markedly reduced NK cells and a mildly elevated NKT cell proportion. By integrating literature findings, we aim to provide novel insights for immune monitoring in the clinical management of GD.

Case presentation

A 58-year-old male was admitted to the Department of Hematology, Huaibei People's Hospital, due to incidentally detected splenomegaly and pancytopenia on a routine physical examination. The patient had no significant past medical history, transfusion history, or tumor history, and there was no family history of genetic disease or consanguineous marriage. Written informed consent was obtained from the patient for the publication of this case report and all accompanying clinical and laboratory images.

The patient was alert and oriented with mild pallor; no petechiae, ecchymoses, or other skin lesions were noted. No enlarged superficial lymph nodes were palpable on systemic examination. Sclerae were anicteric, and the nasal and oral mucosa were intact without blood blisters or ulceration. The pharynx was non-erythematous with non-enlarged tonsils. The neck was supple with no sternal tenderness. Bilateral lung auscultation revealed clear breath sounds with no dry or moist rales. The heart rate was 79 beats per minute with a regular rhythm, and no cardiac murmurs were auscultated. The abdomen was soft and non-tender with no rebound tenderness; the liver was not palpable below the costal margin. Severe splenomegaly was noted with a firm consistency (Line I: 18 cm, Line II: 18 cm, Line III: +3 cm), and no shifting dullness was present. No lower extremity edema was observed. Physiological reflexes were intact, and pathological reflexes were absent.

Color Doppler ultrasound of the liver, gallbladder, pancreas, and spleen demonstrated severe splenomegaly with heterogeneous parenchymal echogenicity and no focal lesion; the liver and pancreas were unremarkable in size and echogenicity, and no free peritoneal fluid was detected.

The patient exhibited pancytopenia in peripheral blood with an elevated erythrocyte sedimentation rate (ESR). Additionally, immune function assays revealed reduced serum levels of total protein, globulin, immunoglobulin G (IgG), immunoglobulin A (IgA), complement C3, and complement C4, all below the normal reference range (Table 1). Furthermore, flow cytometric analysis of peripheral blood lymphocyte subsets (Table 2) (gated on CD45+/side scatter-low lymphocytes) showed the following percentages and absolute counts of cells within the lymphocyte population: helper/inducer T lymphocytes (CD4 + T) at 45.70%, suppressor/cytotoxic T lymphocytes (CD8 + T) at 36.72%, and B lymphocytes at 11.42%, all within normal reference ranges. However, both the percentage (4.44%) and absolute count (21 cells/μL) of NK cells (CD3-CD16 + CD56+) were significantly below the normal reference range. Notably, the percentage of NKT cells (CD3 + CD16+CD56+) was 11.64%, with an absolute count of 55 cells/μL. According to data from a multicenter study of healthy Chinese adults, the mean absolute count of NKT cells is 4.16 ± 3.34 cells/μL, with a commonly used clinical reference range of 10–100 cells/μL [3]. The patient's absolute NKT cell count of 55 cells/μL falls within the upper-middle range of normal reference values. Combined with its percentage approaching the upper limit of normal (0.78%-11.71%), this

suggests a trend of relative preservation or even mild elevation of NKT cells against a background of markedly reduced NK cells (Fig. 1).

Table 1
Summary of hematological examination results for the patient

Item	Test Result	Reference Range
WBC (×10⁹/L)	1.99	3.50 ~ 9.50
RBC (×10¹²/L)	3.05	4.30 ~ 5.80
HGB (g/L)	92	130 ~ 175
PLT (×10⁹/L)	31	125 ~ 350
ESR(mm/h)	77	0.00 ~ 15.0
TP(g/L)	52.3	65.0 ~ 85.0
GLB(g/L)	16	20.0 ~ 40.0
IgG(g/L)	3.74	7.0 ~ 16.0
IgA(g/L)	0.48	0.70 ~ 3.80
C3(g/L)	0.63	0.80 ~ 1.85
C4(g/L)	0.14	0.20 ~ 0.40
GCase(nmol/h/ml)	5.92	6.56 ~ 55.1

Table 2
Results of flow cytometric analysis of lymphocyte subsets

Item	Test Result	Reference Range
CD3 + T(%)	82.74	56 ~ 86
CD3 + CD4+T(%)	45.7	33 ~ 58
CD3 + CD8+T(%)	36.72	13 ~ 39
CD3-CD19 + B(%)	11.42	5 ~ 22
CD3-CD16 + CD56+NK(%)	4.44	5 ~ 26
CD3 + CD16+CD56 + NKT(%)	11.64	0.78 ~ 11.71
CD4 + T/CD8 + T ratio	0.24	0.71 ~ 2.78
Lym(/μL)	469	1530 ~ 3700
CD3 + T(/μL)	388	723 ~ 2737
CD3 + CD4+T(/μL)	215	404 ~ 1612
CD3 + CD8+T(/μL)	172	220 ~ 1129
CD3-CD19 + B(/μL)	53	80 ~ 616
CD3-CD16 + CD56+NK(/μL)	21	84 ~ 724
CD3 + CD16+CD56 + NKT(/μL)	55	10 ~ 100

Bone marrow biopsy reveals grade 1 reticulocytosis. The degree of nucleated cell proliferation in the bone marrow is generally normal (hematopoietic area approximately 45%), with a slightly reduced granulocyte-to-erythrocyte ratio. Granulocytes were infrequent, predominantly in the moderately mature stage. Erythroid cells were scarce, predominantly consisting of mid-to-late stage erythroblasts; the number of megakaryocytes was within normal range, with some exhibiting small cell bodies and reduced lobulation; lymphocytes and plasma cells were sparsely distributed; no collagen fiber proliferation was observed in the bone marrow stroma, with histiocytes distributed in large patches.

Immunohistochemistry results show: CD34 in small vessels (+), round nucleated cells occasionally present (+), E-cadherin in multiple clusters (+), CD61 in megakaryocytes (+), occasional mononuclear cells, CD68 in multiple clusters (+), CD163 sparsely scattered (+), CD3 sparsely scattered (+), CD20 occasionally present (+), MPO in scattered small clusters (+). Large-area distribution of histiocytes is observed, suggesting a lipid metabolism disorder.

Bone marrow morphology examination reveals active bone marrow proliferation with an inverted granulocyte-to-erythrocyte ratio. Granulocyte proliferation remains active at 40.5%, with reduced proportions of band and segmented nuclei; cellular morphology across all stages is generally normal. Erythrocyte proliferation is relatively more active at 41.0%, predominantly involving mid- and late-stage

erythroblasts, with mature erythrocytes exhibiting variable sizes. The megakaryocytic lineage showed active proliferation, predominantly consisting of granular megakaryocytes. A total of 64 megakaryocytes per 2.5×2.5 cm section were observed throughout the slide, with scattered platelets rarely seen. Typical Gaucher cells accounted for 9.0% of the cells examined: large cell bodies with small, displaced nuclei, occasionally binucleated, abundant pale blue cytoplasm containing numerous wrinkled, wavy, fibrous-like structures (Fig. 2).

Final diagnosis: (1) clinical manifestations of severe splenomegaly and pancytopenia; (2) bone marrow findings of typical Gaucher cells and extensive histiocytic infiltration; (3) significantly reduced GCase enzymatic activity; (4) a pathogenic homozygous *GBA1* gene mutation in exon 7: NM_000157.4:c.907C > A (p.Leu303Ile) (Fig. 3); and (5) profound immune dysfunction (hypogammaglobulinemia, hypocomplementemia, and perturbed NK/NKT cell subsets). The patient was diagnosed with adult type I GD complicated with immune dysfunction.

Discussion

This report describes a typical adult type I GD case with a homozygous *GBA1* mutation (NM_000157.4:c.907C > A (p.Leu303Ile)) and a unique immunophenotype of significant NK cell depletion and mild NKT cell elevation, which enriches the immune phenotype spectrum of GD and provides new clinical evidence for understanding the immune pathogenesis of GD.

GD is most prevalent in Eastern European Ashkenazi Jews, with a carrier frequency of 6%, versus 0.7–0.8% in non-Jewish populations. Five subtypes of GD have been identified: types I, II, III, the perinatal lethal form, and the cardiovascular form. Type I is the most common and typically non-neurological, while the rarer types II and III involve central nervous system impairment. Type II and the perinatal lethal form are usually fatal in neonates, and type III may cause premature adult death [4–5]. All GD subtypes stem from *GBA1* gene mutations that impair lysosomal GCase activity, leading to toxic glucosylceramide accumulation, primarily in the liver, spleen and bone marrow [6]. Enzyme replacement therapy (ERT), such as intravenous imiglucerase (a recombinant human GCase), is the first-line treatment for type I GD, yielding significant clinical efficacy and marked improvements in quality of life [7].

GD exemplifies the intricate interplay between genetics, biochemistry, and clinical medicine, characterized by systemic lipid accumulation and a clinical spectrum from mild to severe across all age groups [8]. Bone marrow involvement causes hematopoietic suppression, with common splenomegaly-related cytopenias and notable leukopenia (predominantly lymphopenia) and NK cell depletion [9–10]. Massive Gaucher cell infiltration induces plasma cell dysplasia [11], and lyso-GL1 impairs osteoblast function [12].

Abnormal lipid accumulation in GD impairs the mononuclear phagocyte system (MPS) comprising dendritic cells (DCs), Mo, and Mφ with diverse immune roles. Derived from bone marrow hematopoietic stem cells, Mo differentiate into organ-specific Mφ that mediate immune defense, homeostasis, and antigen presentation in innate/adaptive immunity [13–14]. The MPS supports immune responses by

secreting complement components and cytokines, phagocytosing pathogens, and triggering T-cell responses via MHC class II-antigen complexes. Lipid-deposited Mo/Mφ dysfunction in GD induces immune dysregulation, increasing susceptibility to infections and malignancies [15–17].

However, in this GD case, bone marrow and peripheral blood primarily manifested reduced Mo and lymphocyte counts. Lymphocyte immunophenotyping analysis revealed a particularly significant decline in NK cells, accompanied by a mild expansion of the NKT cell subset. MPS and NK cells are closely linked to human immune function. In the context of GD, NK cells serve as critical innate effector cells, their numerical reduction and functional suppression directly compromise immune surveillance, particularly rendering the patient susceptible to opportunistic infections [18]. As core effector cells of the innate immune system, NK cells not only eliminate virus-infected cells and tumor cells through direct cytotoxic effects but also secrete cytokines like IFN-γ and TNF-α to regulate adaptive immune responses [19]. Therefore, the impaired NK cell activity likely represents a key immunological hallmark underlying the patient's immunodeficiency and increased susceptibility to infections and malignancies in the setting of GD.

Additionally, NKT cells represent a unique T-cell subset characterized by the co-expression of T-cell receptors and NK-cell markers [20]. Upon signal stimulation, NKT cells produce specific cytokines and, as antigen-presenting cells, participate in activating DCs to enhance their immune responses [21]. NKT cells also modulate immune responses in autoimmune and inflammatory diseases, and enhance host defense against infections and malignancies [22–23].As key innate-like effector cells with high heterogeneity, NKT cells act as early responders in immune surveillance. They can mediate direct cytotoxicity, secrete chemokines to recruit DCs, and interact with other immune subsets to coordinate protective immune responses. Accordingly, NKT cells exert a critical role in immune surveillance and the recognition and elimination of abnormal cells [24]. Research also indicates that under conditions of increased lipid load, the CD1d-dependent lipid antigen presentation pathway can be activated, inducing NKT cell proliferation and activation [21]. The elevated proportion of NKT cells may be closely associated with the substantial accumulation of lipid antigens in GD. Glucosylceramide and its derivative Lyso-GL1, as endogenous glycolipid molecules, may be captured by CD1d molecules on antigen-presenting cell surfaces and presented to NKT cells. Under the pathological conditions of GD, the persistent presence of these lipid antigens may lead to chronic stimulation and expansion of NKT cells.

The pathophysiological significance of elevated NKT cells in GD remains controversial. On one hand, NKT cells possess potent immunoregulatory functions. Activated NKT cells can enhance Th1-type immune responses by secreting IFN-γ, thereby reinforcing immune surveillance and antimicrobial immunity. Simultaneously, NKT cells can amplify innate immune effects by activating NK cells and DCs [25]. From this perspective, elevated NKT cell levels may represent an adaptive response attempting to compensate for NK cell dysfunction in the setting of GD-related immune dysregulation. Conversely, NKT cells also possess immunosuppressive functions. Certain NKT cell subsets (e.g., CD4 + NKT cells) can suppress excessive inflammatory responses by secreting Th2 cytokines such as IL-4 and IL-13 or by inducing the expansion of regulatory T cells [22]. Thus, abnormal NKT cell expansion may be a double-

edged sword: moderate NKT cell activation helps maintain protective immunity against infection and inflammation, while excessive expansion may contribute to immune dysregulation, chronic inflammation, or impaired immune surveillance in GD. Therefore, whether a high NKT cell proportion confers protective effects or indicates immune dysfunction requires further functional studies and warrants close clinical attention during the diagnosis and treatment of GD patients.

Thus, for confirmed GD patients, comprehensive immune function assessment is recommended, with particular emphasis on NK and NKT cell counts and ratios. Patients with markedly reduced NK cells should be vigilant for recurrent infections and increased malignancy risk, and prophylactic antimicrobial measures and regular tumor screening may be considered when necessary. For patients with elevated NKT cells, deeper immune function evaluation may be warranted to clarify their functional status and pathophysiological role in GD progression.

Conclusion

We report a case of adult-onset type I GD exhibiting a unique immunophenotype characterized by NK cell depletion with mild NKT cell elevation. This distinctive immunological profile offers new insights into the pathogenesis of immune dysregulation in GD, suggesting that immune abnormalities in GD patients extend beyond the mononuclear-macrophage system to involve broader immune cell lineages. Given the increased risk of infections and malignancies in GD patients, comprehensive immunological assessment (including detailed lymphocyte subset analysis) should be incorporated into clinical management to guide individualized treatment and risk evaluation. Future cohort studies with larger sample sizes are needed to elucidate the clinical significance of NKT cell alterations in GD and explore potential immunomodulatory therapeutic strategies.

Declarations

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Author Contribution

Q.L. : conceptualization, data curation (patient clinical and laboratory data collection), investigation, project administration, resources (patient consent), validation, visualization (tables, figures, CARE Checklist), writing-original draft, writing-review & editing. Q.L. is the corresponding author and takes responsibility for the integrity of the work as a whole.L.W. : investigation, data curation (laboratory testing including complete blood count, biochemical assays), writing-review & editing.L.G. : investigation, data curation (immune phenotyping, flow cytometry analysis of lymphocyte subsets, including NK and NKT cells), writing- review & editing.L.Z. : validation, writing-review & editing.All authors have read and

approved the final version of the manuscript and agree to be accountable for all aspects of the work, ensuring that questions related to the accuracy or integrity of any part are appropriately investigated and resolved.

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Figures

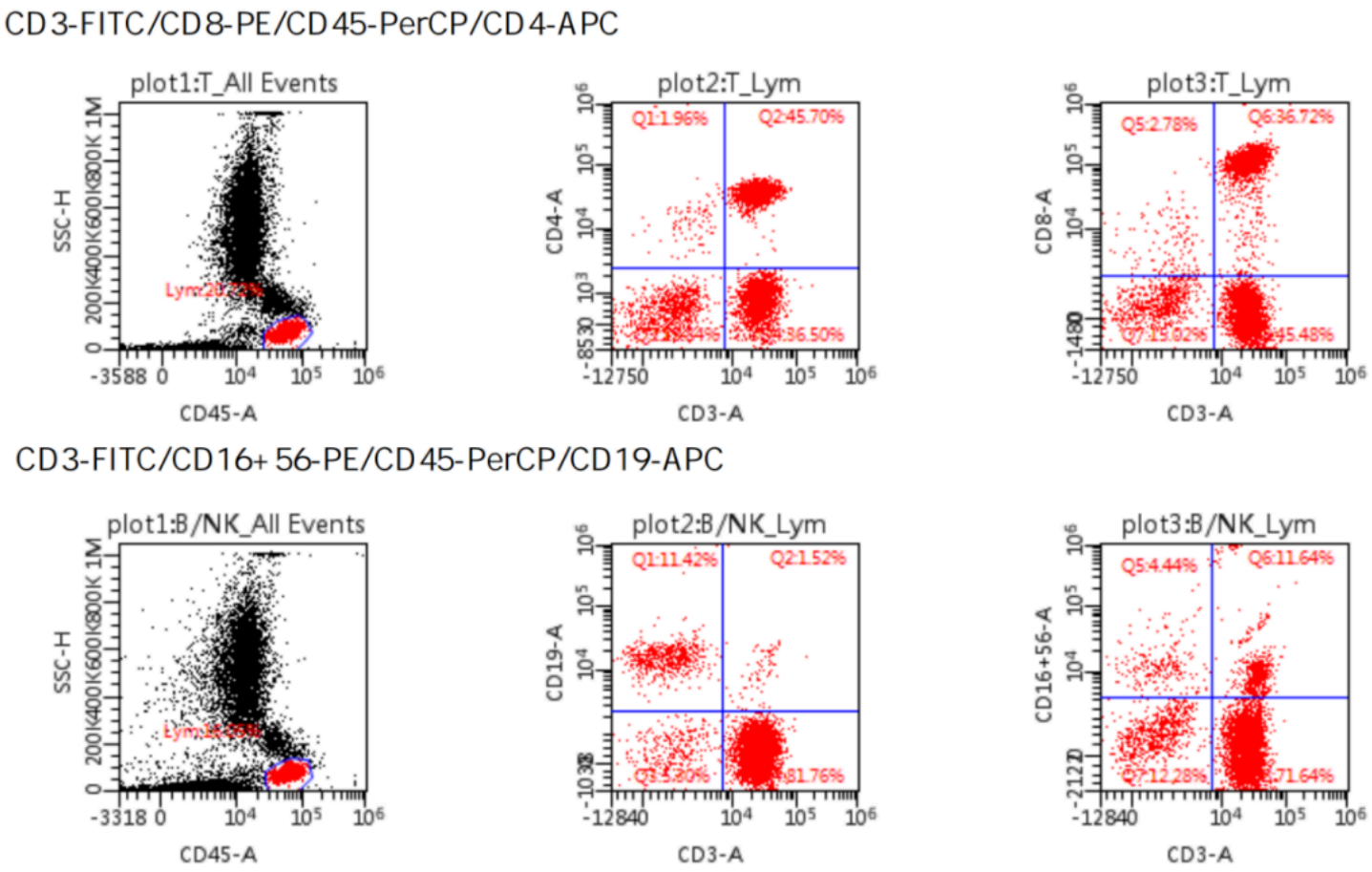


Figure 1

Peripheral blood lymphocytes were analyzed by flow cytometry using antibodies against CD3, CD4, CD8, CD16, CD56, and CD45.

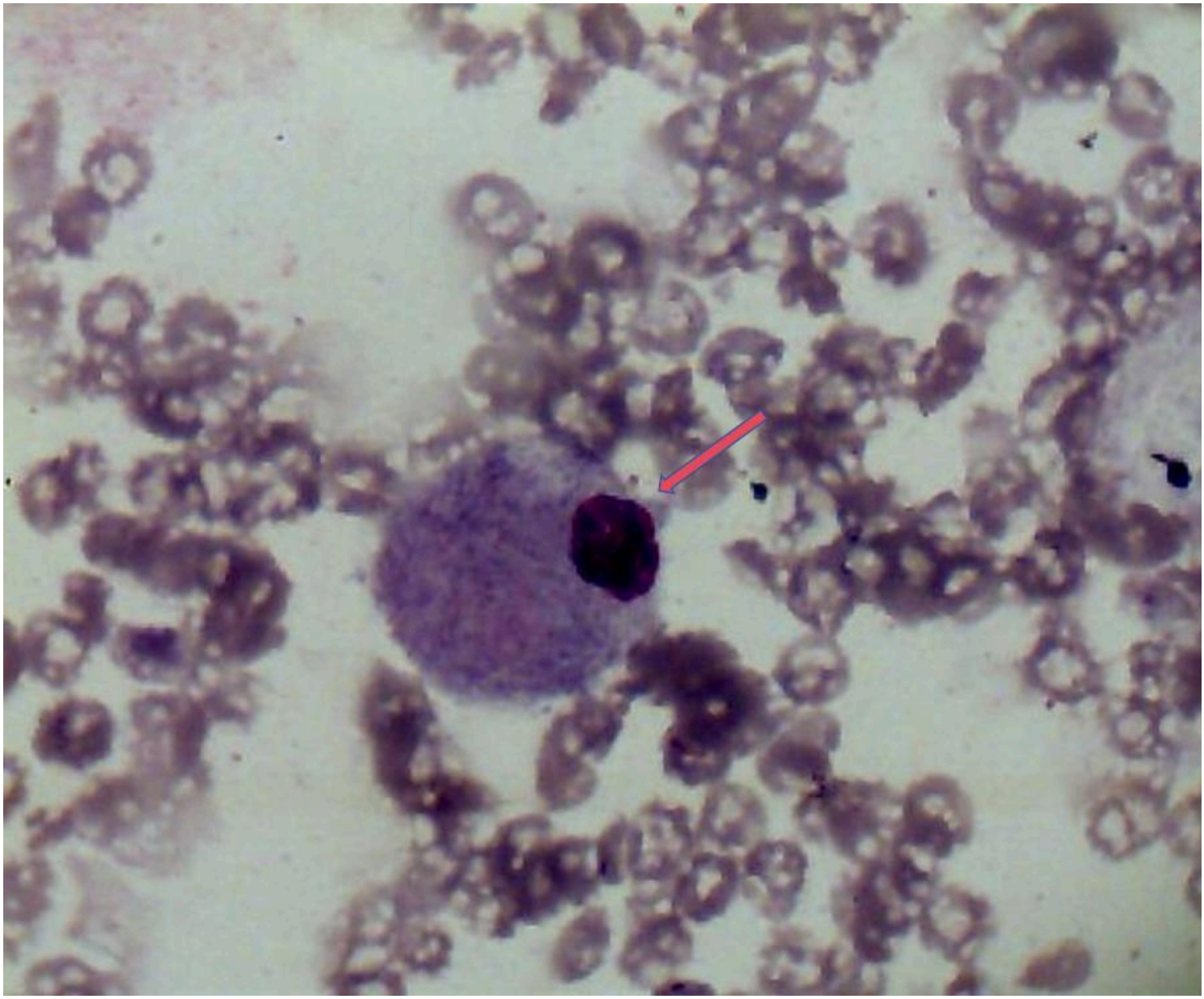


Figure 2

Bone marrow cell morphology (×1000), the red arrow indicates gaucher cell, which is characterized by a large cell body with an eccentrically placed nucleus (occasionally binucleated). The abundant pale blue cytoplasm has a distinctive fibrillary, "crumpled tissue paper" appearance.

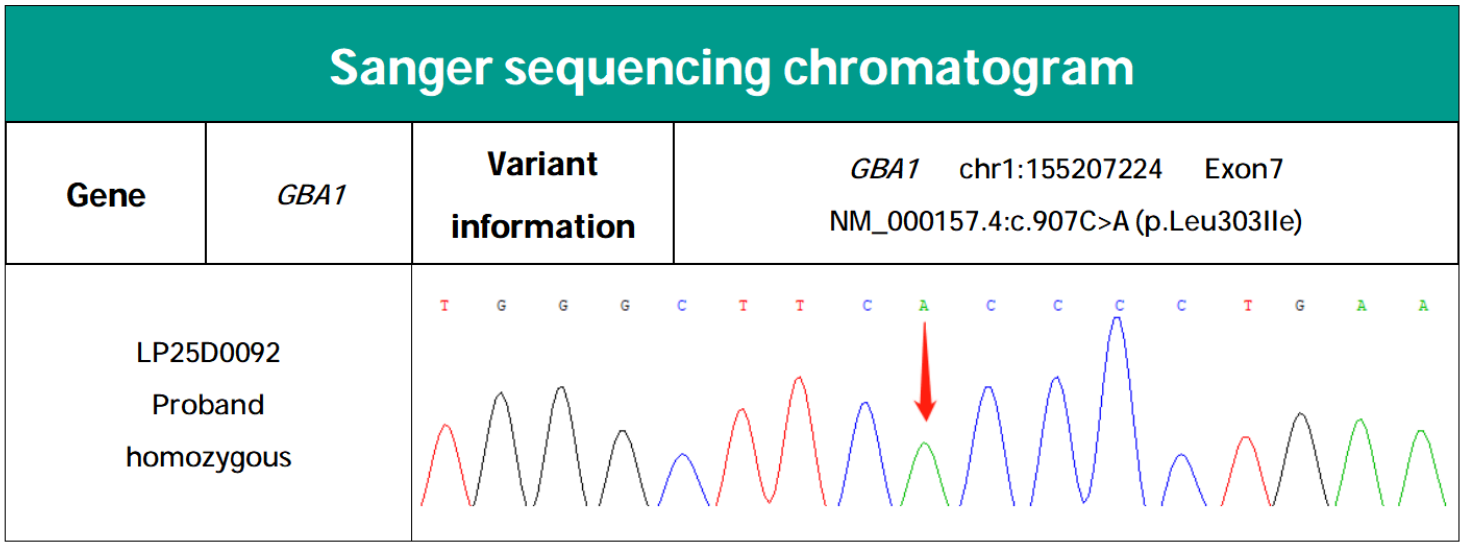


Figure 3

Sanger sequencing chromatogram of the *GBA1* gene (exon 7). The red arrow indicates the homozygous c.907C>A mutation, resulting in a p.Leu303Ile amino acid substitution. The wild-type sequence (C) is replaced by mutant (A) in both alleles.