

Review Article

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Gaucher Disease: Accessible Care is Within Reach

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ABSTRACT

Gaucher Disease (GD) is a rare, potentially life-threatening lysosomal storage disorder caused by mutations in the *GBA1* gene, resulting in deficient β -glucocerebrosidase activity and multisystem involvement. While current therapies offer efficacy for a subset of patients, their prohibitive costs limit accessibility. Although conventional biomedicine attributes GD primarily to genetic and biochemical abnormalities, our previous studies of genetic, congenital, infantile, and early-childhood disorders suggest that these conditions may also have karmic dimensions. Within the Dharma framework, such disorders are understood as manifestations of unresolved karma from previous lives that may be addressed through Dharma practice. Here, we present, to our knowledge, the first case report of a four-year-old girl with biopsy-confirmed GD who, after failure of hematopoietic stem cell transplantation and being considered terminal, subsequently demonstrated remarkable clinical recovery. Her recovery included successful splenectomy without hemorrhagic complications, discharge from sterile isolation, and a progressive return to normal daily activities following her mother's systematic practice of the Three Golden Buddhist Practices—making vows, reciting Buddhist scriptures, and performing life liberation—within the Guan Yin Citta Dharma Door. The patient's preference for sea animals may suggest a pattern of karmic generation that, together with past-life and ancestral karma, may have contributed to the manifestation of her disease. This case challenges an exclusively materialist interpretation of disease and suggests that, within the Dharma framework, addressing proposed karmic factors through Dharma practice may represent a complementary perspective for understanding and supporting healing in rare genetic and early-childhood disorders.

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Introduction

Gaucher Disease (GD) is a rare inherited lysosomal storage disorder caused by pathogenic variants in the *GBA1* gene, resulting in deficient activity of the lysosomal enzyme β -glucocerebrosidase. The consequent accumulation of glucosylceramide and related sphingolipids, particularly within macrophages, leads to the progressive involvement of multiple organ systems. GD is inherited in an autosomal recessive manner and represents one of the most prevalent lysosomal storage disorders, although its incidence varies substantially among populations [1].

The pathophysiology of GD extends beyond the simple accumulation of lipid-laden macrophages, known as Gaucher cells. Dysregulated sphingolipid metabolism, chronic inflammation, immune activation, and secondary effects on bone and hematopoietic tissues contribute to disease progression and clinical manifestations [2,3].

The therapeutic landscape of GD has evolved substantially over the past several decades. Enzyme Replacement Therapy (ERT) with recombinant glucocerebrosidase is an established treatment for many patients and can substantially improve hematologic and visceral manifestations and reduce disease-associated complications [4,5]. Substrate Reduction Therapy

(SRT) provides an alternative or complementary pharmacological strategy by reducing the synthesis of glucosylceramide and thereby decreasing substrate accumulation [6,7]. Beyond ERT and SRT, the broader therapeutic armamentarium now includes pharmacological chaperone therapy, allogeneic hematopoietic stem cell transplantation, emerging gene therapies, and symptom-specific adjunctive care.

Despite these advances, substantial unmet clinical needs remain, particularly for patients with neuronopathic phenotypes, severe skeletal pathology, treatment intolerance, or sub-optimal therapeutic responses [8]. Furthermore, marked heterogeneity persists in clinical severity, genotype-phenotype correlations, and long-term outcomes. Beyond clinical efficacy, the high cost of these disease-modifying therapies renders them inaccessible to many patients, particularly in low- and middle-income countries [9]. Together, these challenges underscore the critical need for novel, cost-effective strategies in GD management.

Our previous studies suggest that genetic diseases may possess underlying karmic and spiritual dimensions, often linked to spiritual attachment [10-12]. In those frameworks, the resolution of karmic debt and spiritual ascension were associated with significant clinical improvement. In the present study, we evaluate whether these Dharma-based principles apply to the etiology and clinical course of GD.

Worldviews, Mechanisms & Solutions

A fundamental divergence between scientific and Dharma-based perspectives lies in their understanding of the nature of life. From a scientific standpoint, life is constituted by biochemical and molecular components, with thoughts and consciousness typically viewed as emergent properties of neural activity within the brain.

In contrast, the Dharma tradition posits that life encompasses both a physical body and a soul or spiritual consciousness that can exist independently of the body under certain conditions, such as at the time of death. Upon separation from the body, this soul may be referred to as a ghost or, more respectfully, a spirit [10-12].

While material possessions and social recognition from one life cannot be carried into the next, karma persists. Any karma that has not been fully retributed in the present life accompanies the soul into subsequent existences, where it manifests itself as suffering through karmic retribution. Genetic diseases and early-childhood disorders are considered typical examples of such retribution from previous lives—a concept supported by prior observations of conditions such as genetic diseases of glutaric aciduria type I, Prader-Willi syndrome, Down syndrome, male genetic infertility, congenital hearing loss, infantile disease of eczema, and early-childhood disease of asthma [13-19].

To deepen our understanding of the Dharma framework surrounding genetic diseases and early-childhood disorders, we will draw on an enlightenment shared by Dharma Master Jun Hong Lu to illustrate this perspective. Because GD is a rare disease, no comparable dialogue or Q&A is available. Therefore, we use a Q&A on congenital glaucoma as an example to illustrate the root causes of these types of disorders.

Q&A 1: Karmic Causes and Retribution for Congenital Glaucoma [20]

(Dialogue took place over the phone on Jan 19, 2016)

Caller: Hello, Master Lu. Could you please take a look at someone born in 1965, in the Year of the Snake? I would like you to check my eyes.

Master: This is quite serious. The retinas at the back of both your eyes have already ruptured.

Caller: Yes, yes. I have congenital glaucoma.

Master: Yes. Let me tell you, the food you eat is not appropriate, and there is something wrong with your blood. This has caused your retinas to rupture. You must stop eating living creatures. You need to make a vow to practise a vegetarian diet.

Caller: Oh, oh, okay.

Master: If you continue eating this way, I am telling you, both of your eyes will be completely ruined.

Caller: Yes, the doctor said that I was already blind in one eye.

Master: Yes.

Caller: And the other eye is also...

Master: It is almost gone as well. I can see that the other eye is in much the same condition. So, you must now make a great vow.

Caller: How many Little Houses would I need to recite?

Master: Start by reciting 180 Little Houses. You must also make a vow to practise a vegetarian diet.

Caller: Okay, okay.

Master: If you do not make the vow to practise a vegetarian diet, I cannot help you. Even reciting the Little Houses will not be effective.

Caller: I see. So, I should first recite 180 Little Houses. What should I do after that?

Master: After that, recite them in batches of 21. You must also make a vow to practise life liberation.

Caller: How many fish should I release?

Master: At least 3,500 fish.

Caller: Okay. Master, is this considered a karmic illness or a spirit-related illness?

Master: A karmic illness.

Caller: I see. It may be because my ancestors were fishermen.

Master: Yes. Because your ancestors caught fish, this is therefore a karmic illness...

This dialogue reveals a striking difference between the Dharma Master's and a medical doctor's approaches to diagnosing the underlying cause of disease. A medical doctor may attribute congenital glaucoma to genetic factors, with surgical intervention remaining the primary definitive treatment, supplemented by supportive medical therapy [21,22]. In contrast, Master Lu identifies karma as the deeper cause of the caller's congenital glaucoma, including his current-life killing of marine animals, his karma from previous lives, and the karmic consequences inherited from his ancestors.

From the Dharma perspective, the first step is to stop creating new karma by making a vow to refrain from eating living creatures. The next step is to eliminate accumulated karma through the Three Golden Buddhist Practices: making vows, reciting Buddhist scriptures (including Little Houses), and performing life liberation. Although Master Lu does not prescribe medical treatment, His teaching emphasizes that addressing and eliminating the underlying karma is essential to resolving the problem at its root.

We propose that GD operates under the same karmic mechanism as other genetic diseases and early-childhood disorders. Accordingly, we hypothesize that by addressing and removing the underlying karmic causes, the expression of such genetic conditions may be ameliorated, offering affected children the potential to lead normal lives.

The method for removing karma is applying the Three Golden Buddhist Practices of Guan Yin Citta Dharma Door in accordance with the principles described previously [10,11].

Results

The following is a sharing presentation by a practitioner of the Guan Yin Citta Dharma Door.

Case 1: Gaucher Disease, a Worldwide Rare Condition, Miraculously Improves Through the Three Golden Buddhist Practices of the Guan Yin Citta Dharma Door

Hello everyone! I am from southeastern China, the mother of two children. My oldest son is 13, and my youngest daughter just turned 4. In June last year, my little daughter suddenly developed many small red spots on her inner thighs. Because she loves to eat sea animals, we initially thought it was a sea animal allergy and did not pay much attention. We kept applying topical skin medications. But the condition recurred persistently and was later misdiagnosed as diaper rash; continued medication brought no improvement.

Then one day in August, I took her to a children's hospital for a check-up. The dermatologist said her symptoms did not look like a skin condition but might be related to a blood disorder. A blood test was done and her platelet count was only $75 \times 10^9/L$ (normal range: $150 - 400 \times 10^9/L$). The doctor referred us to the hematology department of another hospital.

After admission, a series of tests revealed that not only were her platelets problematic, but she also had an enlarged liver and spleen. Her spleen, in particular, was already larger than that of an adult. The doctor advised us to transfer to yet another hospital immediately.

At the third hospital, she underwent blood tests, bone marrow aspiration, genetic testing, and more. At the end of September, she was diagnosed with an extremely rare disease—GD, something we had never even heard of. The hospital explicitly told us they had no medication and that we would have to find it ourselves. Hearing this, I nearly collapsed.

I frantically searched online for information. After a few days, I was in despair. Without treatment, this disease not only causes hepatosplenomegaly and low platelet counts but can also lead to frequent nosebleeds, easy fractures, even disability, convulsions, impaired eye movement, and life-threatening complications. If treated, the only medication is produced in the United States, and it is exorbitantly expensive: one 400-unit vial costs CNY 23,000 (approximately USD 3,410). For my daughter's weight of 13 kg, she would need two vials a month. As she grows, the annual cost for an adult would be around CNY 2 million (approximately USD 290,590), and it requires lifelong administration. Stopping the drug would worsen the condition. How could an ordinary family like ours afford such staggering expenses? I could not bear to think about it and simply could not accept this reality.

On October 21, my husband and I took our daughter to a prestigious hospital in Beijing to consult experts, and the diagnosis was confirmed again. She indeed had GD. Despair struck once more!

A few days later, after calming down, I learned online that there were two other Gaucher patients in two cities in our province. Through a patient support group, I contacted them and visited their homes to learn about their children's conditions. One of those children was already unable to care for themselves.

So, my husband continued working, and I closed my weight-loss shop, which I had run for many years, and began the journey of seeking treatment for my daughter. I heard that traditional Chinese medicine (TCM) might help, so I took her to see a TCM practitioner. After nearly four months of medication, sometimes she took up to seven doses of Chinese and Western medicines a day, yet there was no improvement whatsoever!

In desperation, I turned to friends in the media. Since it was the only known case in our region, in March this year, major newspapers, TV stations, and online outlets reported on her condition. Fortunately, through the coverage, we received help from many kind people. By the end of March, we were able to start her on the medication—the medicine imported from the USA. But the high cost still worried us—if health insurance didn't cover it, how could we sustain it in the future?

I began contacting other GD patients one by one to learn more. Eventually, I found out that some patients had been cured through hematopoietic stem cell transplantation, but the risks were extremely high, even life-threatening. Caught between two difficult choices, we decided that rather than letting her run out of medication later due to cost, we would take a chance on this sliver of hope.

After matching, my husband's bone marrow was compatible for transplantation. So, after more than five months of medication, in July this year, we took her to a hospital in Beijing for a hematopoietic stem cell transplant.

About a month after the transplant, she developed hypersplenism. Her spleen was visibly enlarging day by day. She ran a fever continuously for over a month. The newly engrafted cells kept dropping daily, almost to zero. The doctors tried every medication available but were helpless. She was poked with needles 5 to 8 times a day; almost all her veins were destroyed, to the point that routine venous blood draws were impossible. They had to draw from the arteries. She was covered in bruises.

Even so, several conversations with doctors pushed us to the brink of despair. They told us repeatedly that her death was only a matter of time! Every day I held my daughter and wept in utter hopelessness. I even thought of ending our suffering by jumping from the hospital building, but thinking of my elderly parents and my eldest son, whom I had already neglected, I lacked the courage to do it!

I had never been a devout Buddhist and did not understand karmic retribution. I always believed that as long as I did good deeds, spoke kind words, and helped those around me, that was enough. At home, we had a Buddhist altar, but I often forgot to offer incense. Yet during those desperate days, I could only hold my daughter and chant "Amitābha," silently praying for her to get better. At that time, I did not know about making vows or releasing lives—I simply prayed in my helplessness.

A few days later, we got in touch with Dr. L from a child's Institute, who was willing to perform a splenectomy on her. For an ordinary person, this is routine surgery, but for my daughter, in the doctor's own words: "It is a last-ditch effort."

At that point, she had just undergone a transplant and had no clotting function. Performing surgery with a platelet count of only $2 \times 10^9/L$ meant she could bleed uncontrollably and not survive the operation. I even bought her a new set of clothes, preparing for the worst. When she went into surgery, I could not think clearly—I could only repeat "Namo Guan Yin Bodhisattva" helplessly, with tears streaming down my face.

Grateful to the Bodhisattva for answering our prayers—miraculously, she did not bleed during surgery. It went exceptionally smoothly. The doctor even managed to preserve a portion of her spleen for function. I do not know where the inspiration came from, but right outside the operating room, I made a vow: "from now on, I will eat a vegetarian diet on the 1st and 15th of each lunar month." It was completely spontaneous, yet it came to me naturally. Now I realize that it was the Bodhisattva's blessing that brought me into contact with the Dharma. Deeply grateful to the Greatly Merciful and Greatly Compassionate Guan Yin Bodhisattva!

After surgery, my daughter spent one day in the ICU. Following a blood transfusion, her blood count rose quickly, completely reversing her previous failure to rise. After leaving the ICU, she was transferred back to the regular ward. Her life was saved, but the bone marrow from her father had been metabolized by her spleen. This meant the transplant had failed. We were back to square one, continuing to take medication and applying for health insurance coverage.

A few days later, a friend of over ten years introduced me to a fellow Buddhist practitioner of the Guan Yin Citta Dharma Door! With the careful guidance of that practitioner, I came to know this practice and our master. I gradually understood what karma and karmic obstacles are, and that they can be dissolved through making vows, releasing lives, and reciting sutras and mantras.

I made the following vows:

- Release 50,000 fish over three years for my daughter;
- Recite 600 Little Houses for her karmic creditors;
- Eat a vegetarian diet at least six days a week;
- Once she recovers, I will share her story to inspire others.

Grateful to the Greatly Merciful and Greatly Compassionate Guan Yin Bodhisattva for answering our prayers and extending merciful salvation. After releasing 2,000 fish for her, the very next day she was moved from the sterile ward to a regular ward. We saw hope at once. We continued to release another 2,000 fish twice more. Miracles happen again. The doctors told us she had met the discharge criteria, and could have her intravenous line removed. Once her wound healed, she could go home to recover gradually.

Hearing this, tears of both sorrow and joy poured out like a dam breaking. No more daily needles and endless medication in the hospital. In less than a month, such an incredible, earth-shattering change had occurred. It made me deeply feel the boundless power of the Bodhisattva and the truth of the Dharma. Once again, I thank Namo Greatly Merciful and Greatly Compassionate Guan Yin Bodhisattva who rescues the afflicted!

Now, my daughter is improving day by day, her spirits are rising—she sings, dances, laughs, pesters me for stories, and draws! Watching her get better, I intensively recited the *Great Compassion Mantra* and the *Mantra to Untie Karmic Knots*, making the most of the limited time in the hospital. When I first managed to recite the *Great Compassion Mantra* 49 times, I felt filled with strength and was as happy as a child! Remember, at the beginning I could not even finish one recitation. Also, after eating a vegetarian diet for over half a month, my chronic constipation completely resolved, and I felt physically and mentally refreshed!

Grateful to the Greatly Merciful and Greatly Compassionate Guan Yin Bodhisattva for blessing and protecting us, and to all the fellow Buddhist practitioners who helped us. In the future, I will continue to practice diligently on this path of Buddhist study to repay Buddha's kindness.

With gratitude.
Shared by: Z214

Discussion

GD exemplifies the complex challenges inherent in managing rare, inherited metabolic disorders. While significant therapeutic advances, most notably ERT and SRT, have transformed the prognosis for many patients with type 1 GD, substantial obstacles remain [4,5,7]. These include the high cost and lifelong dependency on therapy, inadequate responses in neuronopathic and severe skeletal phenotypes, and persistent heterogeneity in clinical outcomes [1,3]. The case presented in this study offers a unique perspective by suggesting that clinical improvement in GD may not be solely attributable to biomedical interventions, but may also involve spiritual and karmic dimensions as understood within the Guan Yin Citta Dharma Door framework.

The patient in Case 1 presented with classic, life-threatening manifestations of GD, including profound thrombocytopenia (platelets at $75 \times 10^9/L$), hepatosplenomegaly with splenic enlargement exceeding adult size, and failure of hematopoietic stem cell transplantation due to hypersplenism and graft failure. From a conventional medical standpoint, her survival was precarious, with multiple clinician's warnings indicating that death was imminent. The subsequent recovery—marked by a successful splenectomy without hemorrhagic complications, discharge from the sterile ward, and progressive clinical improvement—occurred in parallel with the initiation of specific Buddhist practices: making vows, performing life release, and reciting assigned Buddhist scriptures, particularly the *Great Compassion Mantra* and *Mantra to Untie Karmic Knots*.

Within the Dharma framework, GD—like other genetic and congenital disorders—is understood as a manifestation of karmic retribution from previous lives, wherein unresolved negative karma accompanies the soul into subsequent existences and materializes as physical disease [10-12]. The practice of the Guan Yin Citta Dharma Door is designed to address these karmic roots through Three Golden Buddhist Practices: making vows, recitation of Buddhist scriptures, and performing life liberations. According to this worldview, the dissolution of karmic obstacles can alleviate the spiritual causes of disease, thereby facilitating physical recovery. The mother's spontaneous vow to adopt a vegetarian diet, followed by her structured vows to release 50,000 fish, recite 600 Little Houses, and share her daughter's story upon recovery, exemplify this transformational process.

The patient's mother did not seek a totem reading from Master Lu; therefore, the precise karmic source of her daughter's condition was not disclosed through that channel. Nevertheless, from the mother's narrative, several karmic factors can be inferred. The family's diet included a substantial quantity of sea animals, indicating a pattern of killing and consumption that generates negative karma. In Dharma understanding, a child born with karma from previous lives is drawn into a family whose existing karmic conditions—particularly those of the parents—serve as an invitation or karmic resonance for such rebirth.

In this case, the young patient not only inherited the karmic residue from her prior existence but also developed a strong preference for consuming sea animals in her current life, thereby generating significant new negative karma. This accumulated new karma, which overlaps upon the pre-existing karmic debt from both her own past lives and her ancestral lineage, is understood to have reached a critical threshold and erupted, manifesting as the severe phenotype of GD.

This constitutes the deepest etiological mechanism—a dimension that remains entirely unexplored by conventional biomedical science, which limits its inquiry to genetic mutations and biochemical pathways. From the Dharma perspective, the genetic defect is not the ultimate cause but rather the physical expression of a karmic condition that operates at the spiritual level, awaiting resolution through appropriate spiritual practice.

However, it is critical to emphasize that the observed clinical improvements occur alongside, and not in replacement for, conventional medical treatments, including ERT, hematopoietic stem cell transplantation, and splenectomy. The case does not support the abandonment of evidence-based therapies; rather, it suggests that spiritual practice may serve as a complementary factor that enhances resilience, facilitates coping, and potentially

optimizes the physiological milieu for recovery. The mother's own report of resolution of chronic constipation and improved mental clarity following dietary changes and recitation practice further underscores the bidirectional relationship among mind, spirit, and physical health.

The broader implication of this case, together with our previous reports on glutaric aciduria type I, Prader-Willi syndrome, Down syndrome, genetic infertility, congenital hearing loss, early-childhood disorders of eczema, and asthma [13-19], is that genetic diseases may share a common etiological layer beyond the molecular mutations identified by genomic medicine. While the *GBA1* pathogenic variant provides the necessary biochemical substrate for GD, the Dharma perspective posits that the expression and clinical trajectory of such diseases are modulated by karmic conditions that can be addressed through spiritual cultivation. This does not negate the validity of genetics, but rather expands the explanatory model to include dimensions currently outside the scope of conventional biomedical research.

The consistency of recovery patterns observed across multiple genetic conditions in our prior work lends some support to the hypothesis that Dharma-based practices may have a meaningful role in improving outcomes for patients with rare diseases, particularly when conventional therapies are inaccessible, prohibitively expensive, or incompletely effective. The fact that the mother's daughter met discharge criteria and began to thrive—singing, dancing, and drawing—after being declared terminally ill, represents a clinically noteworthy event that warrants further investigation.

Future research should aim to collect larger case series with prospective documentation of clinical parameters, standardized spiritual practice protocols, and, where feasible, biomarker assessments (e.g., glucocerebrosidase activity, chitotriosidase, and cytokines) before and after intervention. Collaborative efforts between Dharma practitioners and medical researchers could yield valuable insights into the potential psychospiritual determinants of the disease trajectory, while respecting the epistemological boundaries of both traditions.

Conclusion

This case report documents the first account of clinical improvement in GD associated with the Three Golden Buddhist Practices of the Guan Yin Citta Dharma Door. A four-year-old patient with life-threatening GD, who had failed hematopoietic stem cell transplantation and was declared terminal, demonstrated remarkable recovery—successful splenectomy without bleeding, discharge from sterile isolation, and return to normal activities—following her mother's initiation of formal Dharma practices: making vows, reciting Buddhist scriptures, and performing life liberation.

Consistent with our previous reports on other genetic disorders and early-childhood diseases, this case supports the view that these types of diseases may possess an underlying karmic etiology beyond their molecular mutations. The *GBA1* pathogenic variant is understood not as the ultimate cause but as the physical expression of karmic conditions—including unresolved past-life karma, ancestral karma, and new negative karma from killing and consuming sea animals. Addressing these karmic roots through Dharma practice may facilitate clinical improvement.

In a word, this case study further demonstrates the validity of Master Lu's teachings regarding the causes of genetic and early-childhood diseases.

Acknowledgments

On Master Jun Hong Lu's blog, numerous healing experiences are documented. For the Chinese website, please refer to (<http://www.lujunhong2or.com>). For the English website, please refer to (<https://guanyincitta.com>). Without exception, these cases bear witness to the truth of the Dharma.

Conflict of Interest

None.

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None.

Ethical Statement

The author did not participate in the study design, treatment procedures, or analysis of the reported outcomes. All the experimental procedures and practices by the presenters were done by themselves independently.

Statement by Translator and Writer

The 1 Q&A and 1 case presentation in the text were translated from Chinese to English based on their intended meaning rather than a word-for-word approach. The remaining portions of the paper were written based on my limited understanding of Guan Yin Citta Dharma Door. If there are any inaccuracies or deviations from the true meaning of the Chinese version, or if the content does not accurately reflect Master Lu's teachings, I sincerely seek forgiveness from the Greatly Merciful and Greatly Compassionate Guan Yin Bodhisattva, all Buddhas and Bodhisattvas, Dharma Protectors, and Master Jun Hong Lu.

Artificial Intelligence (AI) tools, including ChatGPT, Gemini, Grok, and Perplexity, were used to assist with translation from Chinese to English, reference duplication checking, and grammatical refinement of the manuscript. The author reviewed and verified all AI-assisted outputs and takes full responsibility for the final content. The assistance provided by these tools is gratefully acknowledged.

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