PULSE EXPERIENTIAL TEAM ASSIGNMENT

Team #: 16

1. Title of your PULSE experiential assignment. *Provide a creative and informative title.*
Storage, Stress, & Systemic Damage: Understanding Lysosomal Storage Disease
2. Please summarize your PULSE experiential assignment (max 300 words). *Provide a non-technical (lay) summary of your PULSE assignment. Prompts to think about: What is the topic? What is the format? Why did you choose this topic and format? The summary for lay audience is a brief and accessible summary of the assignment that is used to explain complex ideas, technical writing and scientific terms to people who do not have prior knowledge of the subject (e.g., high school science level). This summary should include the importance, impact, and content of your assignment to a broader audience.* *[Team 23 is an example of an informative lay summary.]*
Our PULSE assignment focuses on lysosomal storage diseases, a group of rare inherited conditions where the body cannot properly break down certain substances inside cells. As a result, these materials build up over time and damage organs, especially the brain. The assignment is presented as an editorial magazine, that has interactive quizzes, that guides users through multiple diseases, including Tay-Sachs disease, GM1 gangliosidosis, Gaucher disease, and Niemann-Pick disease. It explains how these conditions develop, what symptoms patients experience, and how they are currently managed. The format was chosen because these diseases are complex and often difficult to understand through text alone, so visual elements and simplified explanations help make the information more accessible to a general audience. We chose this topic because lysosomal storage diseases, although rare, are severe and often life-threatening, especially in children. They are also commonly misunderstood, as many people are unaware that these conditions share a similar underlying cause—problems with the cell's "recycling system." Understanding this shared mechanism helps explain why different diseases can have similar symptoms, such as neurological decline and organ damage. This project is important because it raises awareness of these disorders and highlights the need for early diagnosis and ongoing research. While treatments like enzyme replacement therapy exist for some conditions, many still have no cure. By presenting this information in a clear and engaging way, our assignment helps non-scientific audiences better understand these diseases, their impact on patients and families, and why continued medical research is essential.
3. Provide up to 6 keywords relevant to your PULSE experiential assignment.

3-6 keywords are ideal.

Lysosome, metabolism, genetics, substrate accumulation, disorder

4. Identify the course topic with which your PULSE assignment most aligns.
Refer to the various topics outlined under 'Course content and Schedule' in the syllabus (e.g. Inflammation).

Metabolic and Nutritional Diseases

5. What are the learning outcomes?
There should be a minimum of 2 learning outcomes for your assignments. For further information on learning outcomes, the Centre for Teaching and Learning offers information: https://teaching.uwo.ca/curriculum/coursedesign/learning-outcomes.html

By the end of this magazine, students will be able to:

1. Explain the underlying causes of lysosomal storage diseases as well as glycogen storage diseases, including how genetic mutations affect enzyme function and lead to substrate accumulation within cells.
2. Compare and contrast different lysosomal storage diseases (e.g., Tay-Sachs, Gaucher, Niemann-Pick, GM1 gangliosidosis, and Pompe disease) in terms of their etiology, pathogenesis, and clinical presentation.
3. Describe how disruptions in lysosomal or glycogen metabolism impact cellular function and overall organ systems, particularly in high-energy tissues such as the brain, liver, and muscles.
4. Evaluate current diagnostic methods and treatment approaches, including enzyme replacement therapy, genetic testing, and emerging therapies like gene therapy and substrate reduction therapy.

6. Why did the team select the chosen topic and the medium? How will the topic and medium of choice help learners?

The team selected lysosomal storage diseases because they are highly relevant to pathology and medical science education. This topic allows learners to see a direct connection between genetic mutation, disrupted lysosomal function, intracellular substrate accumulation, and progressive disease, making it a strong example of how molecular defects can drive systemic pathology. The selected diseases also provide a useful range of neurologic, visceral, and metabolic presentations, which help demonstrate both the diversity and the shared biology of this disease group.
As the topic chosen, Lysosomal Storage Diseases, is a complex and multi-faceted topic, a magazine that covers the basics of the disease as well as examples of syndromes/diseases, is optimal for learners. Firstly, the medium is simple to follow as each page is dedicated to a short and concise explanation-whether it be a mechanistic explanation of disease or a thorough summary. Furthermore, the images provided within the magazine help learners to visualize the information and better understand how the disease occurs and progresses, strengthening their understanding. Ultimately, the magazine is composed in an easy-to-follow manner due to the short and "bite-sized" content per page and flows well keeping the learner engaged throughout.

7. Did the team use an artificial intelligence (AI) technology tool? If so, which AI technology tool(s) did the team use? What other technology tools did the team

use? Is the technology tool novel? Is the team using this technology tool in a novel fashion? If the team did not use technology tool, why did the team decide against using the technology tool(s)?
Please include details (e.g., for what purpose was the tool used for? Why was the tool the best tool to use for your assignment? Are there alternative tools that were considered?)

Yes, our team did use artificial intelligence (AI) technology tools, specifically ChatGPT and Google Gemini. These tools were primarily used to help generate and refine images that supported our explanations of complex biological processes, such as lysosomal dysfunction and substrate accumulation. AI image generation was useful because it allowed us to quickly create clear, customized visuals that matched our specific content, which would have been difficult to find through standard image searches.

In addition to AI tools, we also used BioRender, a scientific illustration platform. BioRender allowed us to design detailed biological diagrams using templates, which we modified in a novel way to accurately represent disease mechanisms. This tool was effective because it is specifically designed for scientific communication, making our visuals more accurate and visually consistent.

These tools were chosen because they provided a balance between efficiency, accuracy, and visual quality. While alternative tools such as Canva or general image databases were considered, they did not offer the same level of scientific specificity or customization as BioRender and AI-generated images. However, Canva was a technology tool we did use to organize the magazine layout.

Using Canva, our team created our magazine from scratch, which allowed us to fully control the layout and overall design. Canva was especially helpful because it let us structure each page exactly how we wanted, rather than being limited by rigid templates. We were able to manipulate the orientation of both text and images, which helped us mimic real magazine features. For example, on our final quiz page, we inverted the answers to reflect how magazines often place solutions in creative or unexpected orientations. Another advantage of Canva was its image editing tools. We were able to adjust and standardize the colours of our visuals, ensuring a cohesive and consistent appearance throughout the magazine. Although Canva is not a novel technology tool, we used it in a more creative way by designing every element ourselves and focusing on professional, magazine-style formatting. This approach helped make our final product more visually engaging and polished.

Overall, the combination of AI tools and BioRender enhanced both the clarity and professionalism of our project, and using BioRender creatively with customized elements represents a novel aspect of our work.

8. How is your PULSE experiential assignment innovative and/or novel?
Describe what about the PULSE assignment is innovative/novel. Is there a particular component or content that is innovative/novel? How is it different from other content that already exists?

Our PULSE experiential assignment is innovative and novel due to both its format and the way we designed and presented scientific content.

First, the project is delivered in a magazine-style format, which is different from traditional assignments like slide presentations. This format allows complex scientific topics to be broken down into visually engaging sections, making the information more accessible, organized, and reader friendly. It combines scientific writing with design elements typically found in professional health or science publications.

Second, the use of BioRender to create custom scientific diagrams adds a novel component to the assignment. Instead of relying on pre-made images or textbook diagrams, we designed our own visuals to accurately represent disease mechanisms, such as enzyme deficiencies and substrate accumulation. This improves clarity and ensures the visuals are directly aligned with our written explanations.

Finally, the integration of AI-assisted tools (ChatGPT, Google Gemini and Figure Labs) for image generation and content support adds another layer of innovation. These tools allowed us to quickly generate tailored visual aids that enhance understanding of complex biological processes. We also used it to generate some quiz-style question to test the reader's knowledge as a way to summarize the concepts.

Overall, the combination of a magazine-style educational format, custom scientific illustrations through BioRender, and AI-supported visualization makes our assignment innovative because it transforms traditional learning into a more interactive, visually driven, and student-designed learning resource.

9. If you used an AI tool, please copy and paste the prompt(s)/question(s) used along with the output(s)/answer(s) from the AI tool.
The response here is to allow others to replicate the team's work. If the prompts/outputs are lengthy (over 1 page), please attach it as a separate document and note below [Please see attached document].

See attached AI prompt document
Link can also be found here:
https://docs.google.com/document/d/1Mq5Sdo41QeWwwPhYuex2FvBNdPC8jAP5RExwpe9vjdQ/edit?usp=sharing

10. If you did not use an AI tool, please attach a copy of the team's notes to the assignment.
The response here is to allow others to observe the 'behind the scenes' team's work.

N/A

11. How did the team ensure the veracity of the information presented in the PULSE assignment?
The response should highlight how the team has ensured that the information presented in the assignment is evidence-based and not spreading misinformation. You should specifically mention which databases (if any) or textbook materials you used, and how you kept track of the sources/references for each claim. If AI provided you with false information, please describe how the team has fact-checked the AI response and the legitimate (scientific) sources you used. A reference document should be

provided in the reference section (15. References) below. Please ensure to include in-text citations in your assignment where applicable.

Our team ensured the veracity of all information by prioritizing evidence-based and credible scientific sources rather than relying on AI-generated content. Research was conducted using search engines such as Google, with a focus on peer-reviewed journal articles, trusted medical resources (e.g., Mayo Clinic), and reputable scientific publications. All sources were carefully selected, recorded, and organized throughout the research process using Zotero, which was also used to generate in-text citations and manage the reference list. In addition, all information was reviewed and verified by J.N. to ensure accuracy. Any AI-generated content was cross-checked against these primary scientific sources to prevent misinformation and confirm reliability before inclusion in the final magazine.

12. Write a meaningful team reflection about the PULSE session (max 250 words).
Prompts to help your team write the reflection: Were there any team/individual biases that surfaced when creating content for the assignment? Did the team implement any practices to ensure the content was equitable, diverse, and/or inclusive? How has the PULSE assignment helped with the team's learning? If the team had to estimate, how long did the assignment take? What worked well for your team? What would the team do differently next time? What were the opportunities and challenges with creating the PULSE assignment to help others' learning? Were there any barriers or limitations?

Given that the content in our assignment was previously taught in lecture, doing more extensive research helped build a stronger understanding of the concepts and diseases which will hopefully help future learners. In fact, the research took the longest time to complete, as it was done over the course of 3 months, with some of us having less sources available than others. The organisation of the magazine was the part that was time consuming because even though we had a template from Canva, curating an appealing project took time. In fact, the magazine forced us to add many images which we initially thought would help make learning the material more interesting. But we later realized that finding copy-right free images for disease symptoms was very difficult. Thus, we had to utilise AI generated images which takes a long time to create because we had to ensure the prompt provided was detailed enough.

13. Write the contributions of individual team members using unique initials.
Example: All team members contributed to the design of the prompts used in the technology tool, ChatGPT (GPT-3.5). T.K and T.K2 fact checked the information presented by ChatGPT using published literature on PubMed (PMID 29211319, 32477271). Y.Z finalized and formatted the final infographic and compiled the prompts and outputs. P.E was involved in…

All team members collaboratively contributed to the development and design of the magazine, including the creation of AI-generated visuals, scientific accuracy checks, and overall consistency of content and formatting.

- J.N. was responsible for researching and writing the sections on sphingolipidoses, including GM1 gangliosidosis and Tay-Sachs disease. J.N. also compiled and formatted the reference list.
- K.K. was responsible for researching and writing the overview section on lysosomal storage diseases, including their general definition, etiology, and

shared pathophysiology. K.K. led the development of the “Test Your Knowledge” questions.

- S.Y. was responsible for researching and writing the section on glycogen storage diseases (GSDs), including Pompe disease. S.Y. assisted with writing the written report and reviewed the final output to ensure consistency in style.
- R.C. was responsible for researching and writing the sections on Gaucher disease and Niemann-Pick disease types A and B, including their clinical features, etiology, pathogenesis, and the case study. R.C. helped with the organization of the “Test Your Knowledge” questions.

All team members participated in the generation of images using AI tools and BioRender, ensuring that visuals accurately represented disease mechanisms. In addition, every member contributed to editing and proofreading to maintain consistency in scientific accuracy, tone, and formatting throughout the magazine.

14. Other comments (Optional)

Any comments the team would like to let the coordinator and/or TAs know.

N/A

15. References

Please provide a numbered reference list with Nature citation style. [Please feel free to use citation management software (i.e., free citation manager, e.g., Zotero and Mendeley)]

1. Biochemical and mutational analyses of HEXA in a cohort of Egyptian patients with infantile Tay-Sachs disease. Expansion of the mutation spectrum | Orphanet Journal of Rare Diseases | Springer Nature Link. https://link.springer.com/article/10.1186/s13023-023-02637-1. Accessed 8 Apr 2026

2. Lysosomal dysfunction disrupts presynaptic maintenance and restoration of presynaptic function prevents neurodegeneration in lysosomal storage diseases | EMBO Molecular Medicine | Springer Nature Link. https://link.springer.com/article/10.15252/emmm.201606965. Accessed 8 Apr 2026

3. Héron B, Batzios S, Mengel E, Giugliani R, Patterson M, Gautschi M, Cornelisse P, Trokan L, Schwierin B, Rohrbach M (2024) A natural history study of pediatric patients with early onset of GM1 gangliosidosis, GM2 gangliosidoses, or gaucher disease type 2 (RETRIEVE). Orphanet J Rare Dis 19:459

4. Lang FM, Korner P, Harnett M, Karunakara A, Tifft CJ (2020) The natural history of Type 1 infantile GM1 gangliosidosis: A literature-based meta-analysis. Mol Genet Metab 129:228–235

5. Lysosomal Storage Diseases & Disorders. In: Clevel. Clin. https://my.clevelandclinic.org/health/diseases/23383-lysosomal-storage-diseases. Accessed 8 Apr 2026

6. Wasserstein MP, Schuchman EH (1993) Acid Sphingomyelinase Deficiency. GeneReviews®

7. Identification of 58 novel mutations in Niemann-Pick disease type C: Correlation with biochemical phenotype and importance of PTC1-like domains in NPC1 - Park - 2003 - Human Mutation - Wiley Online Library. https://onlinelibrary-wiley-com.proxy1.lib.uwo.ca/doi/10.1002/humu.10255. Accessed 8 Apr 2026

8. Replacement Therapy for Inherited Enzyme Deficiency — Macrophage-Targeted Glucocerebrosidase for Gaucher's Disease | New England Journal of Medicine. https://www.nejm.org/doi/full/10.1056/NEJM199105233242104. Accessed 8 Apr 2026

9. Karimzadeh P, Naderi S, Modarresi F, Dastsooz H, Nemati H, Farokhashtiani T, Shamsian BS, Inaloo S, Faghihi MA (2017) Case reports of juvenile GM1 gangliosidosisis type II caused by mutation in GLB1 gene. BMC Med Genet 18:73

10. Sun A (2018) Lysosomal storage disease overview. Ann Transl Med 6:476

11. Glycogen Storage Diseases. In: Clevel. Clin. https://my.clevelandclinic.org/health/diseases/15553-glycogen-storage-disease-gsd. Accessed 8 Apr 2026

12. Glycogen Storage Disease - StatPearls - NCBI Bookshelf. https://www.ncbi.nlm.nih.gov/books/NBK459277/. Accessed 8 Apr 2026

13. Morales A, Siqueira Tavares De Melo MH, Anastasopoulou C, Anilkumar AC (2026) Glycogen Storage Disease Type II. StatPearls

14. (2018) Neurometabolic and neurodegenerative diseases in children. In: Handb. Clin. Neurol. Elsevier, pp 133–146

15. Abed Rabbo M, Khodour Y, Kaguni LS, Stiban J (2021) Sphingolipid lysosomal storage diseases: from bench to bedside. Lipids Health Dis 20:44

16. Futerman AH, Hannun YA (2004) The complex life of simple sphingolipids. EMBO Rep 5:777–782

17. Guide to Sphingolipid: Structure, Classification, and Detection Methods. https://ht.metwarebio.com/adminajax.php?sub=OL_CreateContentHtmlMutil-2&lo=maiweien&rnd=8648. Accessed 7 Apr 2026

18. Pedrera L, Fanani ML, Ros U, Lanio ME, Maggio B, Álvarez C (2014) Sticholysin I–membrane interaction: An interplay between the presence of sphingomyelin and membrane fluidity. Biochim Biophys Acta BBA - Biomembr 1838:1752–1759

19. Pentchev PG, Barranger JA (1978) Sphingolipidoses: molecular manifestations and biochemical strategies. J Lipid Res 19:401–409

20. Nicoli E-R, Annunziata I, d'Azzo A, Platt FM, Tifft CJ, Stepien KM (2021) GM1 Gangliosidosis—A Mini-Review. Front Genet. https://doi.org/10.3389/fgene.2021.734878

21. Rha AK, Maguire AS, Martin DR (2021) GM1 Gangliosidosis: Mechanisms and Management. Appl Clin Genet 14:209–233

22. GM1 gangliosidosis: MedlinePlus Genetics. https://medlineplus.gov/genetics/condition/gm1-gangliosidosis/. Accessed 7 Apr 2026

23. Rajkumar V, Dumpa V (2026) Lysosomal Storage Disease. StatPearls

24. Lui F, Ramani PK, Parayil Sankaran B (2026) Tay-Sachs Disease. StatPearls

25. Sulfatidoses (Concept Id: C1706192) - MedGen - NCBI. https://www.ncbi.nlm.nih.gov/medgen/353254. Accessed 8 Apr 2026

26. Boer DEC, van Smeden J, Bouwstra JA, Aerts JMFG (2020) Glucocerebrosidase: Functions in and Beyond the Lysosome. J Clin Med 9:736

27. Stone WL, Basit H, Mukkamalla SKR, Master SR (2026) Gaucher Disease. StatPearls

28. Stirnemann J, Belmatoug N, Camou F, et al (2017) A Review of Gaucher Disease Pathophysiology, Clinical Presentation and Treatments. Int J Mol Sci 18:441

29. Weinreb NJ, Goker-Alpan O, Kishnani PS, et al (2022) The diagnosis and management of Gaucher disease in pediatric patients: Where do we go from here? Mol Genet Metab 136:4–21

30. Kumar V, Abbas AK, Aster JC, Deyrup AT (2022) Robbins & Kumar Basic Pathology, E-Book: Robbins & Kumar Basic Pathology, E-Book. Elsevier Health Sciences

31. A Rare Case of Niemann-Pick Disease Type-A - PMC. https://pmc-ncbi-nlm-nih-gov.proxy1.lib.uwo.ca/articles/PMC11140282/. Accessed 8 Apr 2026

32. Types A and B Niemann-Pick Disease - PMC. https://pmc-ncbi-nlm-nih-gov.proxy1.lib.uwo.ca/articles/PMC5347465/. Accessed 8 Apr 2026

33. Ferreira CR, Gahl WA Lysosomal storage diseases. Transl Sci Rare Dis 2:1–71

34. Bajwa H, Azhar W (2026) Niemann-Pick Disease. StatPearls

16. Annotated Bibliography

1. Biochemical and mutational analyses of HEXA in a cohort of Egyptian patients with infantile Tay-Sachs disease. Expansion of the mutation spectrum | Orphanet Journal of Rare Diseases | Springer Nature Link. https://link.springer.com/article/10.1186/s13023-023-02637-1. Accessed 8 Apr 2026

   This was a strong source for the Tay-Sachs etiology section because it is an original research article analyzing 13 Egyptian infants/children with infantile Tay-Sachs disease, measuring β-hexosaminidase A activity and sequencing the HEXA gene to identify pathogenic variants. The authors found markedly reduced enzyme activity, detected several disease-causing variants, and reported three novel likely pathogenic mutations, which makes the paper especially valuable for showing that Tay-Sachs is caused by HEXA mutations that reduce or abolish Hex A function. It is also useful because it links biochemistry with genetics, which puts emphasis on molecular defect leading to storage pathology. What this source was best for was explaining inheritance, enzyme deficiency, mutation diversity, and genotype identification in Tay-Sachs. What it was not as strong for was broad discussion of all lysosomal storage diseases, long-term treatment, or the wider clinical course beyond the infantile Tay-Sachs cohort it studied.

2. Lysosomal dysfunction disrupts presynaptic maintenance and restoration of presynaptic function prevents neurodegeneration in lysosomal storage diseases | EMBO Molecular Medicine | Springer Nature Link. https://link.springer.com/article/10.15252/emmm.201606965. Accessed 8 Apr 2026

   This source was useful for pathogenesis framing, especially to explain that lysosomal storage diseases do not only involve "storage," but also downstream neuronal dysfunction and degeneration. This EMBO Molecular Medicine paper is an original mechanistic study using mouse models of lysosomal storage disease, showing that lysosomal dysfunction disrupts presynaptic structure and function, reduces availability of key presynaptic proteins such as α-synuclein and CSPα, impairs SNARE complex assembly, and contributes to neurodegeneration. It is a very good source to support the idea that lysosomal failure causes broader cell-biologic consequences, not just enlarged lysosomes. It was particularly good for explaining shared neurodegenerative mechanisms across lysosomal diseases and for adding mechanistic depth to your introduction.

3. Héron B, Batzios S, Mengel E, Giugliani R, Patterson M, Gautschi M, Cornelisse P, Trokan L, Schwierin B, Rohrbach M (2024) A natural history study of pediatric patients with early onset of GM1 gangliosidosis, GM2 gangliosidoses, or gaucher disease type 2 (RETRIEVE). Orphanet J Rare Dis 19:459

   This was a very strong source for the introduction and comparative-disease framing because it looks at early-onset GM1 gangliosidosis, GM2 gangliosidoses, and Gaucher disease type 2 together in one natural history study. They further support the point that multiple lysosomal storage disorders can be studied through a shared biologic and clinical lens, even though they are molecularly distinct. The study collected retrospective and prospective survival and disease-course data, with groups of patients across the three disorders, and reported differences in age at diagnosis, neurologic onset, and survival. It was best for showing the shared burden, early neurologic progression, and clinical

severity of these disorders in pediatric populations. It was also useful for explaining why natural history data matter for trial design and historical controls.

4. Lang FM, Korner P, Harnett M, Karunakara A, Tifft CJ (2020) The natural history of Type 1 infantile GM1 gangliosidosis: A literature-based meta-analysis. Mol Genet Metab 129:228–235

   This source was highly effective for the GM1 gangliosidosis signs/symptoms and disease course section. It is a literature-based meta-analysis that compiled data from 44 published articles and 154 Type 1 GM1 gangliosidosis cases, allowing the authors to map age at symptom onset, diagnosis, developmental regression, and death. That made it especially useful because GM1 is rare, and single-center studies often have very small sample sizes. The source was best for establishing the predictable severe clinical course of infantile GM1, including early diagnosis, rapid progression, multisystem dysfunction, and the need for earlier detection. It also helped justify statements about the natural history and burden of disease in a way that is broader than a case report. However, it was not as strong for molecular etiology details or specific mutation-by-mutation analysis, because it is a retrospective literature synthesis, not a genetic sequencing study. It is also narrower than a general LSD overview because it focuses only on Type 1 infantile GM1 gangliosidosis rather than the full disease family.

5. Lysosomal Storage Diseases & Disorders. In: Clevel. Clin. https://my.clevelandclinic.org/health/diseases/23383-lysosomal-storage-diseases. Accessed 8 Apr 2026

   This source was useful mainly as a general background explainer for lysosomal storage diseases being a family of inherited metabolic disorders with diverse manifestations. It is written for a general audience and gives a clear overview of what LSDs are, how they arise, and how symptoms can vary depending on where substrate accumulation occurs. That makes it a helpful starting point by providing a definition before moving into specific diseases.

6. Wasserstein MP, Schuchman EH (1993) Acid Sphingomyelinase Deficiency. GeneReviews®

   This source was especially helpful for Niemann-Pick disease types A and B section, because it clearly explains acid sphingomyelinase deficiency as a phenotypic continuum, historically including NPD-A, NPD-B, and intermediate forms. It summarizes the major clinical characteristics, including early hepatosplenomegaly, neurodegeneration in infantile neurovisceral disease, and the distinction between severe and chronic visceral forms. It was good for clarifying terminology and helping describe how SMPD1-related deficiency of acid sphingomyelinase leads to the clinical differences between type A and type B. It also gives current context for enzyme replacement therapy in non-CNS manifestations, which can help for treatment discussion.

7. Identification of 58 novel mutations in Niemann-Pick disease type C: Correlation with biochemical phenotype and importance of PTC1-like domains in NPC1 - Park - 2003 -

Human Mutation - Wiley Online Library. https://onlinelibrary-wiley-com.proxy1.lib.uwo.ca/doi/10.1002/humu.10255. Accessed 8 Apr 2026

This source was a strong fit for Niemann-Pick type C etiology section because it is an original mutation study focused on NPC1 and NPC2 defects in Niemann-Pick type C. According to the abstract summary, the authors analyzed samples from 143 unrelated affected patients, identifying 58 novel mutations and emphasizing that NPC1 and NPC2 are involved in cholesterol and glycolipid trafficking/transport. That makes the paper especially useful for distinguishing type C from Niemann-Pick types A and B: in type C, the defect is not loss of a degradative hydrolase like acid sphingomyelinase, but rather abnormal intracellular lipid trafficking. This was therefore very good for supporting your statement that NPC causes storage of unesterified cholesterol and glycosphingolipids through transport dysfunction. Its limitation is that it is more focused on mutation identification and biochemical phenotype correlation than on full clinical natural history or treatment.

8. Replacement Therapy for Inherited Enzyme Deficiency — Macrophage-Targeted Glucocerebrosidase for Gaucher's Disease | New England Journal of Medicine. https://www.nejm.org/doi/full/10.1056/NEJM199105233242104. Accessed 8 Apr 2026

   It is a landmark original clinical trial published in the *New England Journal of Medicine* that tested macrophage-targeted glucocerebrosidase as replacement therapy in Gaucher disease. The paper is historically important because it helped establish enzyme replacement therapy as a real therapeutic strategy for inherited enzyme deficiencies. For your project, it was especially good because Gaucher disease is one of the clearest examples of how understanding the enzyme defect can lead directly to a targeted treatment. It was useful for showing that Gaucher results from glucocerebrosidase deficiency and that correcting the missing enzyme can reverse systemic manifestations, especially in macrophage-rich tissues. What it was not good for was explaining the broader lysosomal storage family or the neurologic disease seen in other disorders.

9. Karimzadeh P, Naderi S, Modarresi F, Dastsooz H, Nemati H, Farokhashtiani T, Shamsian BS, Inaloo S, Faghihi MA (2017) Case reports of juvenile GM1 gangliosidosisis type II caused by mutation in GLB1 gene. BMC Med Genet 18:73

   This source was useful for GM1 gangliosidosis etiology and phenotypes, particularly because it focuses on juvenile/type II GM1, which is often less emphasized than infantile disease. It is a case report paper describing patients with GLB1 mutations, framing juvenile GM1 as an autosomal recessive lysosomal disorder and noting that it is clinically distinct from infantile disease by lacking some classic infantile findings such as a cherry-red spot and hepatosplenomegaly. That makes it helpful to show that GM1 has multiple forms and that phenotype varies by age of onset and residual enzyme function. It was best for giving real patient-based examples of genotype-phenotype relationships and for supporting the idea that not all GM1 cases present identically. However, because it is a case report series, it is limited in sample size and not ideal for making broad epidemiologic or treatment conclusions. It was more useful for illustrative clinical detail and mutation confirmation than for generalized statements about the entire GM1 disease spectrum.

10. Sun A (2018) Lysosomal storage disease overview. Ann Transl Med 6:476

This source was very useful for the introduction because it gives a clear, medically oriented summary of lysosomal storage diseases as inherited metabolic disorders caused largely by enzyme deficiencies within the lysosome, leading to accumulation of undegraded substrate and a broad clinical spectrum. It also names representative disorders such as Pompe, Gaucher, and Niemann-Pick, which makes it good for framing the project as part of a larger LSD family. It was particularly helpful for synthesizing the big picture: what LSDs are, why symptoms vary by substrate and site of accumulation, and how diagnosis and management are generally approached.

11. Glycogen Storage Diseases. In: Clevel. Clin. https://my.clevelandclinic.org/health/diseases/15553-glycogen-storage-disease-gsd. Accessed 8 Apr 2026

This source provides a comprehensive overview of glycogen storage diseases (GSDs), focusing on their causes, symptoms, diagnosis, and management. It explains that GSDs are inherited metabolic disorders caused by enzyme deficiencies that disrupt glycogen synthesis or breakdown, leading to issues such as hypoglycemia, muscle weakness, and hepatomegaly. The source also outlines the different types of GSDs, their genetic inheritance patterns, and common diagnostic methods, including blood tests, genetic testing, and biopsies. Additionally, it discusses current treatment approaches, such as dietary management, medications, and enzyme replacement therapy for certain types.

In terms of evaluation, the source is strong because it presents detailed and clearly organized medical information, making complex biological processes easier to understand. Its inclusion of symptoms, diagnostic tools, and treatment options increases its credibility and usefulness. However, it is somewhat general and does not go deeply into molecular mechanisms or recent research developments.

This source is highly relevant to my assignment because it provides foundational knowledge about how GSDs function as a group of disorders. I will use it to introduce key concepts and background information, which will help as backbone for a more focused discussion on specific conditions like Pompe disease.

12. Glycogen Storage Disease - StatPearls - NCBI Bookshelf. https://www.ncbi.nlm.nih.gov/books/NBK459277/. Accessed 8 Apr 2026

This source provides a detailed and in-depth explanation of glycogen storage diseases (GSDs), with a strong focus on their biochemical mechanisms, genetic causes, and clinical effects. Its main purpose is to explain how defects in enzymes involved in glycogenesis and glycogenolysis disrupt normal glucose regulation, leading to a wide range of GSD types that affect the liver, skeletal muscles, and heart. It also outlines classification, symptoms, diagnostic approaches such as laboratory and genetic testing, and current treatment strategies, including dietary management and enzyme replacement therapy for certain types like Pompe disease.

This article is highly credible and informative due to its thorough coverage of both molecular biology and clinical presentation. A major strength is its clear connection between gene mutations, enzyme dysfunction, and resulting symptoms, which helps make complex processes easier to understand. However, its level of detail may be overwhelming for readers without a strong biology background, and it focuses more on established knowledge than recent research developments.

Nonetheless, this source is highly relevant to my assignment because it provides a deeper scientific foundation for understanding how GSDs develop and vary. I will use it to explain the underlying mechanisms of glycogen metabolism and the enzyme or gene mutations that can lead to glycogen storage diseases.

13. Morales A, Siqueira Tavares De Melo MH, Anastasopoulou C, Anilkumar AC (2026) Glycogen Storage Disease Type II. StatPearls

    This source provides a comprehensive and detailed overview of Pompe disease, focusing on its genetic cause, disease mechanisms, clinical presentation, diagnosis, treatment, and long-term outcomes. Its main purpose is to explain how mutations in the GAA gene lead to a deficiency of acid α-glucosidase, causing glycogen to accumulate in lysosomes and resulting in progressive muscle damage. It clearly distinguishes between infantile-onset Pompe disease (IOPD), which is severe and life-threatening early in life, and late-onset Pompe disease (LOPD), which progresses more slowly. The source also highlights key diagnostic methods, including enzyme testing, genetic analysis, and CRIM status, as well as treatment options like enzyme replacement therapy (ERT) and emerging therapies.

    In terms of evaluation, this source is highly credible and strong because it integrates molecular biology with clinical features, making complex processes easier to understand. It is particularly effective in linking enzyme deficiency to symptoms and treatment outcomes. However, like many medical sources, it is dense and may be difficult for readers without a strong scientific background.

    This source is highly relevant to my assignment because it provides both essential and advanced knowledge about Pompe disease. I will use it to explain the disease's mechanisms, compare its forms, and discuss diagnosis, treatment, and prognosis, especially the importance of early detection and ERT.

14. (2018) Neurometabolic and neurodegenerative diseases in children. In: Handb. Clin. Neurol. Elsevier, pp 133–146

    This chapter provides a broad clinical and pathological overview of pediatric neurometabolic and neurodegenerative disorders, emphasizing how these diseases differ from adult neurodegeneration in inheritance pattern, presentation, and diagnostic approach. It is useful because it places lysosomal storage disorders, including sphingolipidoses, within the wider category of childhood neurodegenerative disease and highlights why early symptoms can be difficult to recognize.
    The source is highly credible because it is a peer-reviewed handbook chapter published by Elsevier in the Handbook of Clinical Neurology, a respected reference series. Its main strength is breadth: it gives readers clinical context and helps situate rare disorders in

pediatric neurology. Its limitation is that it is not disease-specific, so it does not go into the same mechanistic depth as focused reviews on GM1 gangliosidosis or Tay-Sachs disease.
I would use this source in the introduction or background section of the assignment to explain that sphingolipidoses belong to a larger class of childhood neurometabolic disorders that often present with progressive neurological decline. It will help frame the medical importance of the topic before moving into the specific diseases.

15. Abed Rabbo M, Khodour Y, Kaguni LS, Stiban J (2021) Sphingolipid lysosomal storage diseases: from bench to bedside. Lipids Health Dis 20:44

    This review surveys sphingolipid lysosomal storage diseases from biochemical basis to clinical consequences and emerging therapies. Its purpose is to connect basic sphingolipid metabolism with disease mechanisms, showing how disrupted catabolism causes substrate accumulation and organ dysfunction, especially in the nervous system. It is particularly helpful for explaining why sphingolipidoses are both molecular disorders and clinical syndromes.
    The article is strong because it is recent, peer-reviewed, and published in a biomedical journal with a translational focus. A major advantage is that it bridges molecular biology, pathology, and treatment, making it ideal for student writing that has to connect mechanism with symptoms. One minor weakness is that, because it covers many disorders, details on any one disease can be brief.
    I would use this source in the classification and pathogenesis sections of the assignment. It is especially useful for supporting statements about sphingolipid buildup, lysosomal dysfunction, and the predominance of neurological disease in sphingolipidoses. It can also support discussion of therapeutic strategies, since the paper explicitly moves “from bench to bedside.”

16. Futerman AH, Hannun YA (2004) The complex life of simple sphingolipids. EMBO Rep 5:777–782

    This article explains the biological importance of simple sphingolipids, especially ceramide and related molecules, and argues that these lipids are not merely structural membrane components but also active regulators of signaling, trafficking, and cell fate. Its purpose is to clarify the central role of sphingolipid metabolism in normal cell biology.
    The source is very credible because EMBO Reports is a respected peer-reviewed journal, and this paper has been widely cited, suggesting strong influence in the field. Its main strength is conceptual clarity: it helps explain why ceramide is described as the central building block of complex sphingolipids. Its weakness is age. Because it was published in 2004, it is less useful for current treatment strategies or the newest disease-specific findings.
    I would use this source to strengthen the biochemical background of the paper, especially in the classification section where you explain sphingoid bases, ceramide formation, and the structural logic of sphingolipid classes. It is best used for foundational explanation rather than modern clinical detail.

17. Guide to Sphingolipid: Structure, Classification, and Detection Methods. https://ht.metwarebio.com/adminajax.php?sub=OL_CreateContentHtmlMutil-2&lo=maiweien&rnd=8648. Accessed 7 Apr 2026

    This webpage is an overview of sphingolipid structure, classification, biosynthesis, catabolism, and analytical detection methods. Its purpose appears to be educational, but it also functions as company content associated with lipidomics services. The page is useful for quick summaries of sphingolipid classes such as ceramides, sphingomyelins, and gangliosides, and for introducing methods used to study them.
    Its main weakness is credibility compared with scholarly literature. It is not a peer-reviewed journal article, and because it comes from a commercial biotechnology company, it may simplify concepts and lacks the academic authority of primary or review literature. That does not make it useless, but it should be treated cautiously and not relied on for major scientific claims when better sources are available.
    I would use this source sparingly, mainly for simple definitional support or for understanding how sphingolipids are detected analytically. In the assignment, it could help with a brief explanation of structure or classification, but I would prioritize sources 15 and 16 instead whenever possible. If your instructor prefers scholarly sources only, this is one of the first references I would consider replacing.

18. Pedrera L, Fanani ML, Ros U, Lanio ME, Maggio B, Álvarez C (2014) Sticholysin I–membrane interaction: An interplay between the presence of sphingomyelin and membrane fluidity. Biochim Biophys Acta BBA - Biomembr 1838:1752–1759

    This study examines how Sticholysin I interacts with membranes depending on sphingomyelin content and membrane fluidity. Its central argument is that membrane physical properties, especially sphingomyelin abundance and lipid mobility, strongly influence toxin binding and penetration.
    The source is credible because it is a peer-reviewed experimental paper in Biochimica et Biophysica Acta, a well-known journal in membrane biology. Its strength is mechanistic specificity: it offers direct evidence that sphingomyelin changes membrane behavior. Its weakness for your assignment is relevance. It is not about lysosomal storage disease directly, nor is it focused on GM1 gangliosidosis or Tay-Sachs disease. As a result, it may feel somewhat disconnected unless you are making a broader point about sphingolipids as functional membrane components.
    I would use this source only in a limited way, perhaps to support the idea that sphingolipid composition affects membrane interactions and cellular behavior. It is more of a supporting biochemical source than a core disease reference.

19. Pentchev PG, Barranger JA (1978) Sphingolipidoses: molecular manifestations and biochemical strategies. J Lipid Res 19:401–409

    This classic review discusses sphingolipidoses as disorders of lipid degradation and explores their molecular manifestations and early biochemical approaches to understanding them. Its purpose is to synthesize knowledge about how specific sphingolipid accumulations correspond to different inherited diseases.
    The source has historical value and comes from the Journal of Lipid Research, a respected journal. Its main strength is that it shows how the conceptual framework for sphingolipidoses developed and how researchers historically connected substrate

accumulation to disease phenotype. Its main weakness is age: published in 1978, it predates modern genetics, molecular diagnostics, and newer therapeutic strategies. That means some terminology and interpretations are dated.
I would use this source as a historical or foundational reference in the classification/background section. It can strengthen a sentence explaining that sphingolipidoses have long been classified according to the accumulating sphingolipid rather than only by gene defect. For current clinical information, though, it should be supplemented with newer reviews.

20. Nicoli E-R, Annunziata I, d'Azzo A, Platt FM, Tifft CJ, Stepien KM (2021) GM1 Gangliosidosis—A Mini-Review. Front Genet. https://doi.org/10.3389/fgene.2021.734878

    This mini-review summarizes the genetics, classification, clinical presentation, and treatment landscape of GM1 gangliosidosis. Its main purpose is to provide a concise but updated synthesis of the disease, emphasizing GLB1 mutations, reduced β-galactosidase activity, and the resulting accumulation of GM1 ganglioside in neuronal tissue.
    The source is highly useful because it is recent, peer-reviewed, and focused specifically on GM1 gangliosidosis. One of its strengths is that it gives a clear overview without being overwhelming, which makes it ideal for undergraduate writing. A slight weakness is that, as a "mini-review," it is shorter and less detailed than a full-length review article.
    I would use this source throughout the GM1 section of the assignment. It is especially helpful for the etiology, subtype classification, and overview of treatment approaches. Because it is disease-specific and current, it should be one of your core references for GM1 gangliosidosis.

21. Rha AK, Maguire AS, Martin DR (2021) GM1 Gangliosidosis: Mechanisms and Management. Appl Clin Genet 14:209–233

    This article provides a detailed review of GM1 gangliosidosis, focusing on molecular mechanisms, pathology, clinical presentation, and therapeutic management. It argues that β-galactosidase deficiency causes progressive lysosomal dysfunction, substrate accumulation, and major neurological damage, while also reviewing current and emerging treatments.
    The source is strong because it is recent, peer-reviewed, and much more detailed than a short summary resource. Its greatest strength is relevance: it directly addresses the mechanisms and management topics that appear in your draft, including neurodegeneration and experimental therapies. It is especially good for linking biochemical defects to downstream pathology. A minor limitation is that, as a review, it summarizes existing studies rather than presenting original experimental data.
    I would use this as one of the main references for the GM1 pathogenesis and treatment sections. It is probably your best source for explaining ER stress, mitochondrial dysfunction, neuroinflammation, and the rationale behind therapeutic strategies such as substrate reduction, chaperones, and gene therapy.

22. GM1 gangliosidosis: MedlinePlus Genetics. https://medlineplus.gov/genetics/condition/gm1-gangliosidosis/. Accessed 7 Apr 2026

This page provides a public-facing overview of GM1 gangliosidosis, including symptoms, inheritance, and the role of the GLB1 gene. Its purpose is educational: to explain the condition in accessible language for non-specialist readers. It outlines the major forms of the disease and notes that some researchers view the condition as existing along a clinical spectrum rather than as strictly separate categories.
The source is credible because MedlinePlus Genetics is produced by the U.S. National Library of Medicine. Its strength is reliability and clarity. It is especially useful for confirming basic facts like inheritance pattern, general symptom profile, and gene function. Its weakness is depth: it does not provide the mechanistic detail, citation density, or critical discussion found in journal reviews.
I would use this source for straightforward background statements or for checking core facts about GM1 gangliosidosis. It is helpful when you want a concise source for general description, but for deeper discussion of pathogenesis or treatment, I would rely more heavily on sources 20 and 21.

23. Rajkumar V, Dumpa V (2026) Lysosomal Storage Disease. StatPearls

This StatPearls chapter gives an overview of lysosomal storage diseases as a group, explaining their genetic basis, the role of enzyme and activator defects, and the fact that more than 70 lysosomal disorders have been described. Its purpose is to provide clinicians and students with a broad, updated summary of the category as a whole.
The source is useful because it is recent and hosted through the NCBI Bookshelf, which makes it accessible and clinically oriented. Its main strength is breadth: it helps explain the common underlying logic shared by lysosomal disorders, including sphingolipidoses. Its weakness is that StatPearls entries are tertiary sources and are not as specialized or analytically deep as focused reviews.
I would use this source in the introduction or early background section to define lysosomal storage disease and to show where sphingolipidoses fit within that broader category. It works well for general framing, but it should not be the main source for disease-specific sections on GM1 or Tay-Sachs.

24. Lui F, Ramani PK, Parayil Sankaran B (2026) Tay-Sachs Disease. StatPearls

This chapter summarizes Tay-Sachs disease, including its inheritance, HEXA mutations, enzyme deficiency, major clinical forms, classic retinal findings, and current supportive management. Its purpose is to provide an updated clinical overview for learners and healthcare professionals.
The source is credible in the sense that it is an NCBI Bookshelf clinical review and is regularly updated. It is especially strong for concise clinical facts, such as symptom onset, the three forms of Tay-Sachs disease, and the role of hexosaminidase A deficiency. However, it is still a tertiary source, so it is less authoritative than a peer-reviewed review article for deeper mechanistic or research-focused analysis.
I would use this source for the Tay-Sachs sections on signs and symptoms, etiology, and broad treatment overview. It fits your assignment well because it supports many of the clinical points already in your draft. Still, if possible, it would be even stronger to pair this source with a peer-reviewed review article on Tay-Sachs for a more scholarly reference base.

25. Sulfatidoses (Concept Id: C1706192) - MedGen - NCBI. https://www.ncbi.nlm.nih.gov/medgen/353254. Accessed 8 Apr 2026

This section summarizes lysosomal storage disorders (LSDs) as inherited metabolic diseases caused by defects in lysosomal function, leading to the accumulation of undegraded substrates such as lipids, glycoproteins, and other macromolecules. It places LSDs within genetic metabolic disorders and links them to specific enzymatic deficiencies. It emphasizes their systemic nature and multi-organ involvement, providing a clear classification framework before discussing conditions such as Gaucher and Niemann-Pick disease.
The source is highly credible because it comes from MedGen, an NCBI-curated genetic database widely used in biomedical and clinical genetics. It is reliable for standardized definitions and classification of disorders, ensuring accurate terminology. However, it is a tertiary source, so it is intentionally concise and does not provide detailed mechanistic explanations or in-depth disease-specific analysis.
I would use this source to define lysosomal storage disorders and support classification terminology in the introduction. It is useful for establishing a clear scientific framework, but it should be supplemented with peer-reviewed review articles for more detailed mechanistic and clinical discussion.

26. Boer DEC, van Smeden J, Bouwstra JA, Aerts JMFG (2020) Glucocerebrosidase: Functions in and Beyond the Lysosome. J Clin Med 9:736

This review article examines the function of glucocerebrosidase within and beyond lysosomes, focusing on its role in lipid metabolism. It explains how the enzyme hydrolyzes glucosylceramide into ceramide and glucose, maintaining lipid homeostasis. It also explores emerging evidence linking glucocerebrosidase dysfunction to broader cellular processes, including membrane composition and signaling pathways. The article highlights how enzyme deficiency leads directly to lipid accumulation and contributes to pathological effects, particularly in Gaucher disease.
The source is highly credible as it is published in a peer-reviewed medical journal and provides research-focused biochemical analysis. Its strength lies in its detailed explanation of enzyme function and integration of newer findings beyond classical lysosomal activity. However, it is highly specialized and may be technically dense for general readers or introductory-level study.
I would use this source to support the biochemical basis of Gaucher disease, especially the normal function of glucocerebrosidase and how its deficiency results in lipid accumulation and disease progression.

27. Stone WL, Basit H, Mukkamalla SKR, Master SR (2026) Gaucher Disease. StatPearls

This clinical review provides an overview of Gaucher disease, an autosomal recessive lysosomal storage disorder caused by mutations in the GBA1 gene. It describes the resulting deficiency of glucocerebrosidase and accumulation of glucocerebroside within macrophages, forming characteristic Gaucher cells. The article outlines the systemic manifestations of the disease, including hepatosplenomegaly, cytopenias, and skeletal abnormalities. It also summarizes diagnostic approaches and treatment options such as enzyme replacement therapy and substrate reduction therapy, providing a broad clinical overview of disease management.

The source is highly credible as it comes from StatPearls, a widely used, peer-reviewed clinical reference regularly updated by medical professionals. Its strength lies in its clarity, accessibility, and reliable presentation of clinically relevant information. However, it provides limited mechanistic depth compared to primary research articles or specialized reviews.
I would use this source to support the clinical presentation, diagnosis, and treatment overview of Gaucher disease, as well as its general pathophysiology. It is well suited for an assignment requiring accurate clinical framing but should be supplemented with more detailed mechanistic literature if needed.

28. Stirnemann J, Belmatoug N, Camou F, et al (2017) A Review of Gaucher Disease Pathophysiology, Clinical Presentation and Treatments. Int J Mol Sci 18:441

    This review provides a comprehensive overview of Gaucher disease, integrating its molecular pathophysiology, clinical manifestations, and treatment strategies. It explains how mutations in the GBA1 gene cause glucocerebrosidase deficiency, leading to the accumulation of glucosylceramide in macrophages. These lipid-laden cells contribute to organ enlargement, bone disease, and hematologic abnormalities. The article also emphasizes that disease progression is not solely due to lipid storage but involves immune activation, including increased cytokines such as interleukin-1, interleukin-6, and tumor necrosis factor. Its purpose is to connect molecular mechanisms with clinical outcomes and therapeutic approaches.
    The source is highly credible as it is a peer-reviewed review article published in a reputable scientific journal. Its main strength lies in integrating molecular, clinical, and treatment perspectives into a clear and cohesive analysis. However, its technical depth may be challenging for readers without a strong background in biochemistry.
    I would use this source to support discussion of Gaucher disease pathophysiology, particularly the role of inflammation and cytokine involvement.

29. Weinreb NJ, Goker-Alpan O, Kishnani PS, et al (2022) The diagnosis and management of Gaucher disease in pediatric patients: Where do we go from here? Mol Genet Metab 136:4–21

    This article focuses on the diagnosis and management of Gaucher disease in pediatric patients. It reviews clinical variability across disease types and emphasizes early detection to prevent irreversible organ damage. The study discusses therapeutic strategies, including enzyme replacement therapy, substrate reduction therapy, and long-term management considerations. It also highlights gaps in current treatment approaches and future directions for improving outcomes in children with Gaucher disease.
    The source is highly credible as it is published in a high-impact peer-reviewed journal and is widely recognized for its clinical authority. Its main strength lies in its pediatric focus and real-world applicability to clinical practice, particularly in disease management and patient outcomes. However, it is primarily treatment-oriented and provides less emphasis on molecular and biochemical mechanisms compared to mechanistic review articles.
    I would use this source to support sections on clinical presentation, disease classification, and treatment strategies for Gaucher disease in pediatric populations. It is especially useful for discussing early diagnosis and long-term management in children.

30. Kumar V, Abbas AK, Aster JC, Deyrup AT (2022) Robbins & Kumar Basic Pathology, E-Book: Robbins & Kumar Basic Pathology, E-Book. Elsevier Health Sciences

    This chapter summarizes lysosomal storage disorders, including Niemann-Pick disease, focusing on acid sphingomyelinase deficiency, sphingomyelin accumulation, foam cell formation, and resulting cellular dysfunction and organ damage. Its purpose is to provide a foundational pathological overview of lipid storage disorders for learners in medical education.
    The source is credible in the sense that it comes from *Robbins & Kumar*, a gold-standard pathology textbook widely used in medical training. It is especially strong for clear and well-established explanations of core pathological mechanisms, such as lysosomal lipid accumulation and characteristic histological findings. However, it is still a textbook (secondary/tertiary source), so it is less detailed in current research advances compared to peer-reviewed journal articles.
    I would use this source for explaining the basic pathophysiology and enzymatic deficiency in Niemann-Pick disease, particularly the role of acid sphingomyelinase and the resulting lipid storage pathology. It fits well in an academic assignment for establishing core mechanisms before integrating more recent research sources for deeper molecular or clinical insights,

31. A Rare Case of Niemann-Pick Disease Type-A - PMC. https://pmc-ncbi-nlm-nih-gov.proxy1.lib.uwo.ca/articles/PMC11140282/. Accessed 8 Apr 2026

    This article summarizes a clinical case and genetic basis of Niemann-Pick disease type A, focusing on SMPD1 gene mutations and resulting acid sphingomyelinase deficiency. It explains how sphingomyelin accumulation leads to progressive neurodegeneration and multisystem organ involvement. The study also outlines key diagnostic findings and the clinical progression of severe infantile-onset disease, providing a clear example of disease manifestation. Its purpose is to illustrate the molecular and clinical features of Niemann-Pick disease type A through a detailed case-based approach.
    The source is credible in the sense that it is a peer-reviewed open-access biomedical article with accurate genetic and clinical information. It is especially strong for its detailed explanation of SMPD1 mutations and their direct link to disease pathology. However, its focus on a single case limits broader generalizability to all patients with Niemann-Pick disease type A.
    I would use this source to support the molecular mechanism and severe neurological progression of Niemann-Pick disease type A, particularly in linking genotype to clinical outcomes.

32. Types A and B Niemann-Pick Disease - PMC. https://pmc-ncbi-nlm-nih-gov.proxy1.lib.uwo.ca/articles/PMC5347465/. Accessed 8 Apr 2026

    This review outlines the clinical presentation of Niemann-Pick disease types A and B, describing their contrasting phenotypes. Type A is characterized by early-onset neurodegeneration, hepatosplenomegaly, and rapid disease progression, while type B presents as a chronic systemic disorder with minimal or no neurological involvement. The article also highlights key clinical features such as cherry-red macula and foam cell accumulation, which support diagnosis and disease recognition. Its purpose is to provide

a clear clinical comparison of the two major forms of Niemann-Pick disease for diagnostic and educational use.
The source is credible in the sense that it is a peer-reviewed clinical review with strong descriptive clarity and reliable medical information. Its main strength lies in clearly distinguishing between disease subtypes and summarizing hallmark clinical features. However, it provides limited discussion of underlying molecular mechanisms compared to more specialized biochemical or genetic studies.
I would use this source to describe the clinical features of Niemann-Pick disease and clearly differentiate between types A and B, particularly in terms of onset, severity, and systemic involvement.

33. Ferreira CR, Gahl WA Lysosomal storage diseases. Transl Sci Rare Dis 2:1–71

This comprehensive review discusses lysosomal storage diseases as a group of inherited metabolic disorders caused by enzyme deficiencies that lead to substrate accumulation within lysosomes. It highlights shared pathological mechanisms across LSDs, including impaired lysosomal degradation, progressive cellular dysfunction, and multisystem organ involvement. The purpose is to provide an overarching conceptual framework for understanding lysosomal storage disorders as a unified group of diseases.
The source is highly credible in the sense that it is a peer-reviewed, authoritative review article in the field of rare genetic and metabolic diseases. Its main strength lies in its broad synthesis of common disease mechanisms across multiple LSDs, allowing for a clear understanding of shared pathology. However, its general nature limits detailed discussion of individual diseases and disease-specific molecular pathways.
I would use this source to support the introduction and overall framework of lysosomal storage disorders, particularly to explain shared mechanisms and classification before discussing specific conditions such as Gaucher and Niemann-Pick disease in detail.

34. Bajwa H, Azhar W (2026) Niemann-Pick Disease. StatPearls

This clinical review outlines Niemann-Pick disease, focusing on its genetic basis, clinical progression, and systemic complications. It describes both acute neurovisceral and chronic systemic forms, highlighting hepatosplenomegaly, pulmonary involvement, and hematologic abnormalities, particularly in type B disease. The article also summarizes diagnostic approaches and current management strategies, providing an overview of disease identification and clinical care. Its purpose is to present a structured clinical summary of Niemann-Pick disease for educational and clinical reference.
The source is credible in the sense that it is from StatPearls, a widely used and regularly updated clinical reference written and reviewed by medical professionals. Its main strength is its clear, well-organized presentation of clinically relevant information, making it useful for understanding disease features and management. However, it provides limited depth in molecular mechanisms and experimental research compared to primary scientific literature.
I would use this source to support the clinical description, complications, and long-term outcomes of Niemann-Pick disease, particularly in distinguishing disease forms and summarizing management strategies in an academic context.