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REVIEW

Genetic epidemiology of NLSD, Chanarin Dorfman (NLSD-I) and Neutral Lipid Storage with Myopathy (NLSD-M). A retrospective review

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Neutral Lipid Storage Disease (NLSD) refers to a group of rare, inherited metabolic disorders characterized by the defective degradation of cytoplasmic triglycerides, leading to their accumulation in various tissues. A classic finding in both disorders is Jordan's anomaly, which consists of lipid-filled vacuoles within neutrophils and other leukocytes on a peripheral blood smear; this phenomenon frequently serves as a critical diagnostic clue.

The genetic epidemiology of NLSD is characterized by autosomal recessive transmission, a strong association with consanguinity, and two distinct genetic aetiologies corresponding to different clinical subtypes. However, this epidemiology has not yet been fully investigated.

Chanarin-Dorfman Syndrome (CDS) is inherited as an autosomal recessive disorder. It is extremely rare, with approximately 186 cases reported worldwide. The major findings of CDS are non-bolluos congenital ichthyosiform erythroderma (NCIE), hepatomegaly, and liver steatosis. CDS is most frequently described in populations from Turkey, Mediterranean countries, the Middle East, North Africa, Pakistan, and neighboring regions. A higher prevalence is consistently associated with consanguineous marriages, which increase the likelihood of inheriting two pathogenic *ABHD5* gene variants.

Similarly, Neutral Lipid Storage Disease with Myopathy (NLSDM) is instead inherited as an autosomal recessive disorder, though it is considered even rarer than CDS, with only 142 cases reported globally. To date, cases of NLSDM have been reported across Europe (particularly in Italy), China, Japan, and Middle Eastern populations. Unlike CDS, many NLSDM patients present during adulthood – typically in their 20s to 40s – manifesting with progressive muscle weakness and significantly elevated serum creatine kinase (CK) levels. Furthermore, cardiomyopathy represents a major cause of morbidity in this specific cohort.

Key words: neutral lipid storage disease, chanarin-dorfman syndrome, neutral lipid storage with myopathy

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Introduction

Chanarin–Dorfman Syndrome (CDS), also known as Neutral Lipid Storage Disease with Ichthyosis (NLSDI), and Neutral Lipid Storage Disease with Myopathy (NLSDM) although sharing a common morphological hallmark characterized by intracellular triglyceride accumulation (Tab. I) are genetically distinct entities. CDS is caused by mutations in the *ABHD5* gene (located on chromosome 3p21.33 which encodes the CGI-58 activator protein.

This syndrome, characterized by congenital ichthyosis, multisystem involvement, and Jor-

Table I. Neutral Lipid Storage Disease Classification.

Chanarin-Dorfman syndrome (NLSDI)	***ABHD5* (CGI-58)**	Congenital ichthyosis, liver disease, variable myopathy, hearing and eye involvement
Neutral Lipid Storage Disease with Myopathy	***PNPLA2* (ATGL)**	Progressive skeletal muscle weakness, elevated CK, cardiomyopathy, little or no skin involvement
ABHD5 activates adipose triglyceride lipase (ATGL), while *PNPLA2* encodes ATGL itself. Defects in either pathway impair triglyceride hydrolysis and lead to lipid droplet accumulation in multiple tissues.		

dan's anomaly was described independently by Chanarin and Dorfman. Historically, CDS was first documented in Italy at the Gaslini Institute in a child found to be affected by a CGI-58 (*ABHD5*) defect, a finding subsequently corroborated by other cohorts [1–3]. NLSDI typically presents with an early age of onset, frequently during the first or second decade of life. Conversely, NLSDM is caused by mutations in the *PNPLA2* gene, which encodes adipose triglyceride lipase (ATGL), a key enzyme in lipid catabolism.

These genetic mutations can either disrupt the patatin-like domain of the protein or impair the ATGL–LD interaction. This binding plays a pivotal role in targeting ATGL to lipid droplets (LDs), thereby facilitating their degradation through a coordinated mechanism of lipolysis and lipophagy. Consequently, *PNPLA2* gene defects lead to progressive skeletal and cardiac muscle damage, alongside pathogenic LD accumulation in tissues. Both disorders are inherited as autosomal recessive traits, requiring the inheritance of two pathogenic alleles. Notably, intrafamilial phenotypic heterogeneity can occur; patients harboring identical mutations may present with divergent clinical patterns including distal asymmetric myopathy, elevated creatine kinase (CK) levels, diabetes, and liver disease within the same family [4]. In this context, serum microRNAs as well as the presence of Jordan's anomaly (Fig. 1) have emerged as valuable diagnostic biomarkers [4,5].

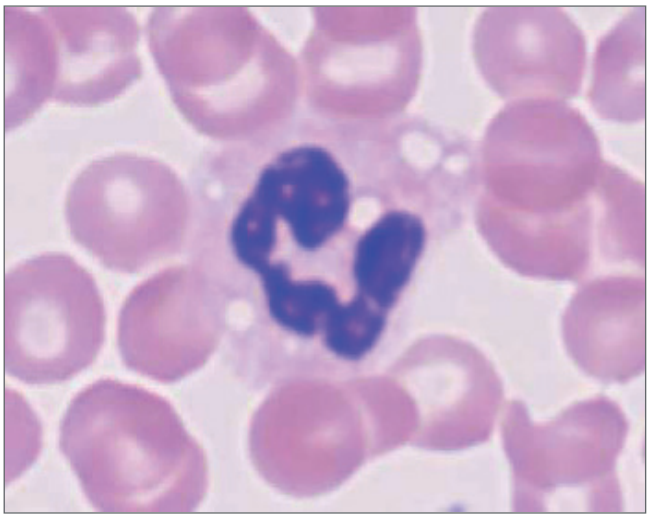


Figure 1. Jordan's anomaly: neutrophil leukocyte shows evidence of leukocyte vacuoles with neutral lipids due to a deficiency in the degradation of cytoplasmic triglycerides, with lipid droplets. Peripheral blood smear (X 400).

Methods

We conducted a retrospective review of molecularly proven NLSDI and NLSDM cases reported worldwide between 1975 and 2025. The investigation focused on characterizing their genetic epidemiology, geographical distribution, country of origin, and the presence of private mutations within patients and families.

Results

CDS. Chanarin–Dorfman syndrome is a rare autosomal recessive (AR) disorder primarily encountered in patients of Mediterranean and Middle Eastern origin (Tab. II). Patients typically come to medical attention due to ichthyosis. We found 130 cases in Mediterranean countries, including 10 cases in Italy, 3 in Greece, and 48 in Turkey or neighboring countries. Two siblings had CNS involvement, myopathy, neurosensory hearing loss, cataracts, nystagmus, strabismus, and mental impairment; their molecular analysis revealed the presence of private variants, specifically the N209* mutation [4]. Patients with ichthyosis or Jordan's anomaly are frequently referred to a muscle clinic for CK elevation, and muscle biopsies are consistent with lipid storage. In Palestine, 5 cases were detected harboring the common mutation c.960+5G > A; furthermore, 9 cases were identified in Israel, 4 in Algeria, 26 in Tunisia, 3 in France, 7 in Spain, 4 in Egypt, and 3 in Morocco, and in India 20 cases, 19 mutations were found. To date, 186 CDS patients have been reported worldwide. NLSDI patients typically have a disease onset in the first or second decade of life. Usually, they are admitted for congenital ichthyosis, Jordan's anomaly, and subsequent liver steatosis. CNS changes may also be detected. The myopathological picture is consistent with lipid storage. Lipid myopathy and liver dysfunction can be treated with a *Medium Chain Triglycerides* (MCT) diet, and retinoids - such as acitretin - can be useful for skin treatment, even in the presence of liver damage.

NLSDM. Mutations in the *PNPLA2* gene, which encodes adipose triglyceride lipase (ATGL), a crucial enzyme for lipid catabolism, cause NLSDM, a disorder characterized by progressive skeletal and cardiac muscle damage and lipid droplet (LD) accumulation in tissues. Mutations in the *PNPLA2* gene might affect the patatin-like domain, while others impair the Adipose Triglyceride Lipase (ATGL) – LD binding hydrophobic domain. Recently, mutations in the microtubule-associated protein 1A/1B-light chain 3 (LC3) domain have been reported. These mutations impair ATGL targeting to LDs, enhancing their degradation through a combined mechanism of lipolysis and lipophagy.

Mutations are inherited as an autosomal recessive trait, wherein a patient inherits two copies of the mutated gene, one from each parent. Phenotypic patterns associated with the same mutation can vary considerably, presenting as distal asymmetric myopathy, elevated

Table IIa. Mutations described in NLSDI cases (186 patients - 130 of Mediterranean origin).

	Country	Number of patients	Mutations
Asia (111 patients)	Afghanistan	3	c.1006G>T
	China	6	c.700C>T; c.838C>T; c.40G>T; c.507-1G>T; c.838C>T; c.461T>G; c.933dupA ; c.99_103del ; c.770C>G
	Fiji	1	ND
	India	20	c.752A>C; c.773+1delG; c.506-3C>G; c.297C>A; c.683T>G; ND
	South-East Asian	1	c.700C>T
	Iraq	4	ND
	Iran	4	c.560_578del19bp
	Israel	10	c.506ins101 bp; c.943C>T; c.700C>T
	Japan	2	c.550C>T; c.215T>C
	Jordan	1	c.588_591delTTGG
	Lebanon	2	c.748T>G
	Palestine	5	c.960+5G>A
	Pakistan	6	c.730_731insA; c.338G>T
	Saudi	2	ND
	Turkey	52	c.778G>A; c.594insC; c.343A>G; c.960+6A>T; c.150C>G; c.217T>G; c.47+1G>A; c.413G>A; c.811G>A; c.29_30del; c.343A>G
	United Arab Emirates	1	c.838C>T
Africa (1 patients)	Uganda	1	ND
North Africa (37 patients)	Algeria	4	c.98C>G; c.389A>C; c.135-2A>G
	Egypt	4	c.700C>T; c.553delTTGGGGTTTCCCT
	Morocco	3	c.19G>A
	Tunisia	26	c.773-1G>A
Europe (25 patients)	Austria	1	c.700C>T
	France	3	c.46_47delAG; c.245A>G
	Germany	1	c.700C>T
	Greece	3	c.898_*320del662; c.1330_773+46del
	Italy	10	c.98C>G; c.890C>T; c.507-1G>A; c.552G>A; c.673delG; c.47+1G>A
	Spain	8	c.943C>T; c.616insGGGdel31bp; c.594insC; c.934G>T c.934G>T
South America (2 patients)	Brazil	1	Large deletion in promoter
	Venezuela	1	Exon 3 deletion

Table IIb. Type of mutation and number of patients by country of origin.

Type of Mutation	Country	Number of patients
Large deletion in promoter	Brazil	1
c.19G>A	Morocco	2
c.29_30del	Turkey	1
c.40G>T;	China	1
c.46_47delAG	Algeria	1
c.47+1G>A	Italy Morocco Turkey	1 1 2
c.98C>G	Algeria Italy	1 1
c.99_103del	China	1
c.135-2A>G	Algeria	2
c.150C>G	Turkey	1
c.215T>C	Japan	1

c.217T>G	Turkey	1
c.245A>G	France	1
c.297C>A	India	1
c.338G>T	Pakistan	1
c.343A>G	Turkey	7
c.389A>C	Algeria	1
c.413G>A	Turkey	1
c.461T>G	China	1
c.506ins101 bp	Israel	4
c.506-3C>G	India	1
c.507-1G>A	Italy	2
c.507-1G>T	China	1
c.550C>T	Japan	1
c.553delTTGGGGTTTCCCT	Egypt	2
c.560_578del19bp	Iran	2
c.588_591delTTGG	Jordan	1
c.594insC	Spain; Turkey	3 28
c.616insGGGdel31bp	Spain	1
c.673delG	Italy	1
c.683T>G	India	1
c.700C>T	Austria China Egypt Germany Israel South-East Asian	1 1 1 1 1 1
c.730_731insA	Pakistan	5
c.748T>G	Lebanon	2
c.752A>C	India	3
c.770C>G	China	1
c.773-1G>A	Tunisia	19
c.773+1delG	India	1
c.778G>A	Turkey	1
c.811G>A	Tyrkey	1
c.838C>T	China; United Arab Emirates	2 1
c.890C>T	Italy	1
c.898_*320del662	Greece	3
c.933dupA	China	1
c.934G>T	Spain	2
c.943C>T	Israel Spain	1 1
c.960+5G>A	Israel Palestine	1 5
c.960+6A>T	Turkey	1
c.1006G>T	Afghanistan	3
c.1330_773+46del	Greece	2
Exon 3 deletion	Venezuela	1

CK, diabetes, and liver disease within the same family [6]. Furthermore, serum microRNAs as well as Jordan's anomaly serve as useful biomarkers [7]. A large cohort of patients has been followed clinically, by genetic analysis, and via imaging in Italy [8,9]. Recently, in a Slovakian case, carrying c.24G>A and c. 798dupC mutations, ATGL was found to be absent on Western blot analysis [11]. Its likely that other mutations could undergo to the same molecular mechanism.

Metabolic treatment is not curative, since no causative therapy is currently available for NLSDM; therefore, clinical management focuses on alleviating symptoms associated with the disease. This typically involves a multidisciplinary approach, including dietary modifications such as an MCT diet, physical therapy, medications to manage symptoms, and regular monitoring of lipid accumulation and organ function. Notably, the MCT diet showed no clinical benefit in a previously tracked patient [9]. Conversely, in patient-derived fibroblasts, triheptanoin treatment induced an increase in glycolysis and mitochondrial respiration compared with controls. These preliminary results indicate that triheptanoin exerts an anaplerotic effect in NLSDM fibroblasts [10] and may pave the way with a repurposed drug for new therapeutic strategies.

Discussion

Neutral Lipid Storage Disease (NLSD) represents a group of rare genetic disorders characterized by the abnormal storage of neutral lipids within various tissues alongside Jordan's anomaly in leukocytes. Globally, NLSD is caused by mutations in two key genes that regulate lipid metabolism, delineating two primary clinical subtypes: NLSD with Myopathy (NLSDM) and NLSD with Ichthyosis (NLSDI). The latter was historically described as a multisystem disorder in a boy [1] who was found to be affected by a CGI-58 (*ABHD5*) defect, a finding subsequently corroborated by other cohorts [2,3].

Table IIIa. Mutations described in NLSDM cases (142 patients).

	Country	Number of patients	Mutations
Asia (102 patients)	China	75	c.187+1G>A; c.749A>C; c.757+2T>C; c.757+1G>T; c.467delC; c.155C>G; c.659_667del; c.353T>C; c.475_478dupCTCC; c.245G>A; c.564G>A; c.333_334del; c.918del; c.749A>C; c.434G>A; c.6_7insT; c.697-1G>A; c.516C>G; c.291_292insT; c.798delC; c.516del; c.922del; c.750_757del; c.757+1G>T; c.863C>G; c.32C>G; c.919delG; exon6-10del; c.132C>G
	Iran	10	c.695delT; c.553T>C; c.32C>T; c.1343dupT; c.613dupC; c.399_404del
	Iraque	1	c.695delT
	Lebanon	2	c.662G>C
	Japan	8	c.475_478dupCTCC; c.799_802delGCCC; c.865C>T; c.696+1G>C; c.576delC; c.194del; c.40G>A
	Pakistan	1	c.497A>G
	Turkey	3	c.613dupC; c.496G>C
	Vietman	2	c.757+1G>T
Europe (26 patients)	Austria	1	c.497A>G
	France	3	c.847delC; c.865C>T; c.798_799insC
	Germany	1	c.1051delC
	Greece	1	c.1081G>T
	Hungary	1	c.24G>A; c.798dupC
	Italy	16	c.541delAC; c.24G>A; c.516C>A; c.424A>T; c.865A>T; c.570A>C; c.497A>G; c.1442C>T; c.177T>G; c.577A>T; c.714C>A; c.41_47delGCTGCGG; c.187+1G>C; c.696+4A>G; c.553_565delGTCCCCCTTCTCG; c.45_47delCGG
	Netherlands	2	c.584C>T; c.808delC; c.584C>T
	UK	1	c.543delC
North Africa (1 patients)	Algeria	1	c.865C>T
North America (2 patients)	Canada	1	c.584C>T; c.1447G>C
	USA	1	retrotrasposon insertion
South America (3 patients)	Brazil	1	c.792del; c.1453A>G
	Chile	1	c.584C>T
	Colombia	1	c.557C>G

Conversely, mutations in the *PNPLA2* gene, which encodes adipose triglyceride lipase (ATGL) – a crucial enzyme for intracellular lipid catabolism – underlie NLSDM. This specific disorder is characterized by progressive skeletal and cardiac muscle damage resulting from pathogenic lipid droplet (LD) accumulation in tissues. Genetic mutations within PNPLA2 might disrupt the patatin-like domain of the protein, while others impair the ATGL–LC3 interaction. This binding plays a pivotal role in targeting ATGL to LDs, thereby enhancing their degradation through a coordinated, combined mechanism of lipolysis and lipophagy.

Both conditions are inherited as an autosomal recessive trait, requiring the patient to inherit two copies of the mutated gene, one from each parent. Notably, intrafamilial phenotypic heterogeneity can occur; patients harboring identical mutations within the same family can present with divergent clinical patterns, including distal asymmetric myopathy, elevated CK, diabetes, and liver disease [6]. For instance, a truncating mutation was shown to lead to a particularly

Table IIIb. Type of mutation and number of patients by country of origin.

Type of Mutation	Country	Number of patients
c.6_7insT	China	1
c.24G>A	Hungary; Italy	1; 2
c.32C>G	China	1
c.32C>T	Iran	2
c.40G>A	Japan	1
c.41_47delGCTGCGG	Italy	1
c.45_47delCGG	Italy	1
c.132C>G	China	1
c.155C>G	China	1
c.177T>G	Italy	3
c.187+1G>A	China	12
c.187+1G>C	Italy	3
c.194del	Japan	1
c.245G>A	China	7
c.291_292insT	China	1
c.333_334del	China	1
c.353T>C	China	1
c.399_404del	Iran	1
c.424A>T	Italy	1
c.434G>A	China	1
c.467delC	China	3
c.475_478dupCTCC	China; Japan	3; 2
c.497A>G	Austria; Italy; Pakistan	1; 1; 1
c.516C>A	Italy	1
c.516C>G	China	3
c.516del	China	1
c.541delAC	Italy	2
c.543delC	UK	1
c.553T>C	Iran	3
c.553_565delGTCCCCCTTCTCG	Italy	1
c.557C>G	Colombia	1
c.564G>A	China	1
c.570A>C	Italy	1
c.576delC	Japan	1
c.577A>T	Italy	3
c.584C>T	Canada; Chile; Netherlands	1; 1; 2
c.613dupC	Iran; Turkey	3; 2
c.659_667del	China	1
c.662G>C	Lebanon	2
c.695delT	Iran; Iraque	1; 1
c.696+1G>C	China; Japan	1; 1
c.696+4A>G	Italy	1
c.697-1G>A	China	1
c.714C>A	Italy	1
c.732_754del TGGCCTGCGCTTTCTGCAGCGGA	China	1
c.749A>C	China	3
c.750_757del	China	1
c.757+1G>T	China; Vietnam	32; 2
c.757+1G>T	China	1
c.757+2T>C	China	3
c.792del	Brazil	1
c.798delC	China	1
c.798_799insC	France; Hungary	1; 1
c.799_802delGCCC	Japan	2
c.808delC	Netherlands	1
c.847delC	France	1
c.863C>G	China	2
c.865A>T	Italy	1

c.865C>T	Algeria; France; Japan	1; 1; 1
c.918del	China	2
c.919delG	China	1
c.922del	China	1
c.1051delC	Germany	1
c.1081G>T	Greece	1
c.1343dupT	Iran	1
c.1442C>T	Italy	1
c.1447G>C	Canada	1
c.1453A>G	Brazil	1
retrotrasposon insertion	USA	1
exon6-10del	China	1

severe, early-onset form in a Slovakian case [10].

Currently, metabolic treatment is not curative, as no gene-targeted or causative therapy is available for NLSD. Consequently, clinical management focuses on alleviating symptoms associated with the disease. This typically requires a multidisciplinary approach, including physical therapy, medical management of complications, regular monitoring of lipid accumulation and organ function, and dietary modifications such as a medium-chain triglyceride (MCT) diet. However, the efficacy of dietary changes remains variable; for example, the MCT diet showed no clinical benefit in a previously tracked patient [9]. Conversely, in vitro studies on patient-derived fibroblasts demonstrated that triheptanoin and a repurposed drug treatment induced a significant increase in both glycolysis and mitochondrial respiration compared with controls. These preliminary results indicate that triheptanoin exerts a powerful anaplerotic effect in NLSDM fibroblasts [11], potentially paving the way for new therapeutic strategies.

Conclusions

Our comprehensive review of Neutral Lipid Storage Disease (NLSD) cases spanning from 1975 to 2025 underscores the distinct genetic epidemiology, geographic clustering, and clinical heterogeneity that characterize both the NLSDI (CDS) and NLSDM subtypes. Despite their shared morphological hallmark of intracellular triglyceride accumulation, their divergent molecular aetiologies - mapped to the *ABHD5* and *PNPLA2* genes respectively - dictate vastly different clinical trajectories. Identifying non-invasive diagnostic clues, such as Jordan's anomaly on peripheral blood smears and emerging serum microRNA profiles, remains paramount for achieving early diagnosis and preventing extensive systemic or muscular damage. Furthermore, the higher prevalence of these disorders in specific geographical regions highlights the profound impact of consanguinity on their genetic transmission and emphasizes the need for targeted genetic screening and counselling in affected populations. Because current therapeutic options are largely symptomatic and standard dietary interventions like the MCT diet often yield suboptimal results in NLSDM, expanding our therapeutic arsenal is critical. In this scenario, the promising anaplerotic effects demonstrated by triheptanoin and repurposed drug trials in patient-derived fibroblasts provide a strong rationale for future clinical trials. Ultimately, establishing international multi-centric registries will be essential to better characterize the natural history of these rare diseases and to accelerate the development of targeted, curative therapies.

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Conflict of interest statement

The authors declare no conflict of interest.

Authors contribution

CA writing and presenting at ICNMD in Florence; DT, SM, and EM mutation data collection.

Ethical consideration

Ethical approval and informed consent were not required for this study as it is based exclusively on previously published data and does not involve new research on human participants.

References

1 Angelini C, Philippart M, Borrone C, et al. Multisystem triglyceride storage disorder with impaired long-chain fatty acid oxidation. Ann Neurol. 1980 Jan;7(1):5-10. https://doi.org/10.1002/ana.410070104.

2 Bruno C, Bertini E, Di Rocco M, et al. Clinical and genetic characterization of Chanarin-Dorfman syndrome. Biochem Biophys Res Commun. 2008 May 16;369(4):1125-8. https://doi.org/10.1016/j.bbrc.2008.03.010.

3 Lefèvre C, Jobard F, Caux F, et al. Mutations in CGI-58, the gene encoding a new protein of the esterase/lipase/thioesterase subfamily, in Chanarin-Dorfman syndrome. Am J Hum Genet. 2001 Nov;69(5):1002-12. https://doi.org/10.1086/324121.

4 Tavian D, Durdu M, Angelini C, et al. Recurrent N209* ABHD5 mutation in two unreported families with Chanarin Dorfman Syndrome..Eur J Transl Myol. 2021 May 12; 31(2):9796. https://doi.org/10.4081/ejtm.2021.9796.

5 Missaglia S, Tasca E, Angelini C, et al. Novel missense mutations in PNPLA2 causing late onset and clinical heterogeneity of neutral lipid storage disease with myopathy in three siblings. Mol Genet Metab. 2015 Jun-Jul; 115(2-3):110-7.

https://doi.org/10.1016/j.ymgme.2015.05.001.

6 Pegoraro V, Missaglia S, Marozzo R, et al. MiRNAs as biomarkers of phenotype in neutral lipid storage disease with myopathy.Muscle Nerve. 2020 Feb;61(2):253-257. https://doi.org/10.1002/mus.26761.

7 Pennisi EM, Arca M, Bertini E, et al; Italian NLSD Group. Neutral Lipid Storage Diseases: clinical/genetic features and natural history in a large cohort of Italian patients. Orphanet J Rare Dis. 2017 May 12;1 2(1):90. https://doi.org/10.1186/s13023-017-0646-9.

8 Garibaldi M, Tasca G, Diaz-Manera J, et al. Muscle MRI in neutral lipid storage disease (NLSD). J Neurol. 2017 Jul; 264(7):1334-1342. https://doi.org/10.1007/s00415-017-8498-8.

9 Pegoraro V, Missaglia S, Marozzo R, et al. Neutral lipid storage disease with myopathy: A 10-year follow-up case report. Eur J Transl Myol. 2022 Jun 17; 32(2):10645. https://doi.org/10.4081/ejtm.2022.10645.

10 Missaglia S, Martegani E, Angelini C, et al. Case Report: Pathogenic PNPLA2 variants and nonsense-mediated mRNA decay result in an early-onset neutral lipid storage disease with myopathy. Front Genet. 2025 Aug 21; 16:1642442. https://doi.org/10.3389/fgene.2025.1642442.

11 Noguera NI, Tavian D, Angelini C, et al. Effects of Triheptanoin on Mitochondrial Respiration and Glycolysis in Cultured Fibroblasts from Neutral Lipid Storage Disease Type M (NLSD-M) Patients. Biomolecules. 2023 Mar 1; 13(3):452. https://doi.org/10.3390/biom13030452.