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Reference List

- Adams D. Recent advances in the treatment of familial amyloid polyneuropathy. *Ther Adv Neurol Disord*. 2013;6(2):129-139. doi:10.1177/1756285612470192
- Adams D, Gonzalez-Duarte A, O'Riordan WD, et al. Patisiran, an RNAi therapeutic, for hereditary transthyretin amyloidosis. *N Engl J Med*. 2018;379(1):11-21. doi:10.1056/NEJMoa1716153
- Adams D, Algalarrondo V, Polydefkis M, Sarswat N, Slama MS, Nativi-Nicolau J. Expert opinion on monitoring symptomatic hereditary transthyretin-mediated amyloidosis and assessment of disease progression. *Orphanet J Rare Dis*. 2021;16(1):411. doi:10.1186/s13023-021-01960-9
- Adams D, Ando Y, Beirao JM, et al. Expert consensus recommendations to improve diagnosis of ATTR amyloidosis with polyneuropathy. *J Neurol*. 2021;268(6):2109-2122. doi:10.1007/s00415-019-09688-0
- Adams D, Tournev IL, Taylor MS, et al. Efficacy and safety of vutrisiran for patients with hereditary transthyretin-mediated amyloidosis with polyneuropathy: a randomized clinical trial. *Amyloid*. 2023;30(1):1-9. doi:10.1080/13506129.2022.2091985
- Adams D, Wixner J, Polydefkis M, et al. Five-year results with patisiran for hereditary transthyretin amyloidosis with polyneuropathy: a randomized clinical trial with open-label extension. *JAMA Neurol*. 2025;82(3):228-236. doi:10.1001/jamaneurol.2024.4631
- Alnylam Pharmaceuticals. Alnylam announces FDA approval of AMVUTTRA® (vutrisiran), the first RNAi therapeutic to reduce cardiovascular death, hospitalizations and urgent heart failure visits in adults with ATTR amyloidosis with cardiomyopathy (ATTR-CM). Published March 20, 2025. Accessed April 3, 2025. <https://investors.alnylam.com/press-release?id=28831>
- AMVUTTRA. Prescribing information. Alnylam; 2025. Accessed March 21, 2025. https://www.accessdata.fda.gov/drugsatfda_docs/label/2025/215515s005lbl.pdf
- Ando Y, Adams D, Benson MD, et al. Guidelines and new directions in the therapy and monitoring of ATTRv amyloidosis. *Amyloid*. 2022;29(3):143-155. doi:10.1080/13506129.2022.2052838
- ATTRUBY. Prescribing information. BridgeBio; 2024. Accessed March 20, 2025. https://www.accessdata.fda.gov/drugsatfda_docs/label/2024/216540s000lbl.pdf
- Benson MD, Dasgupta NR, Rao R. Diagnosis and screening of patients with hereditary transthyretin amyloidosis (hATTR): current strategies and guidelines. *Ther Clin Risk Manag*. 2020;16:749-758. doi:10.2147/TCRM.S185677
- Benson MD. Liver transplantation and transthyretin amyloidosis. *Muscle Nerve*. 2013;47(2):157-162. doi:10.1002/mus.23521
- Berk JL, Suhr OB, Obici L, et al. Repurposing diflunisal for familial amyloid polyneuropathy: a randomized clinical trial. *JAMA*. 2013;310(24):2658-2667. doi:10.1001/jama.2013.283815



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- Bishop E, Brown EE, Fajardo J, Barouch LA, Judge DP, Halushka MK. Seven factors predict a delayed diagnosis of cardiac amyloidosis. *Amyloid*. 2018;25(3):174-179. doi:10.1080/13506129.2018.1498782
- Brito D, Albrecht FC, de Arenaza DP, et al. World Heart Federation consensus on transthyretin amyloidosis cardiomyopathy (ATTR-CM). *Glob Heart*. 2023;18(1):59. doi:10.5334/gh.1262
- Bumma N, Kahwash R, Parikh SV, et al. Multidisciplinary amyloidosis care in the era of personalized medicine. *Front Neurol*. 2022;13:935936. doi:10.3389/fneur.2022.935936
- Casasnovas C, Llado L, Borrachero C, et al. A narrative review and expert recommendations on the assessment of the clinical manifestations, follow-up, and management of post-OLT patients with ATTRv amyloidosis. *Ther Adv Neurol Disord*. 2023;16:17562864231191590. doi:10.1177/17562864231191590
- Coelho T, Marques W, Jr., Dasgupta NR, et al. Eplontersen for hereditary transthyretin amyloidosis with polyneuropathy. *JAMA*. 2023;330(15):1448-1458. doi:10.1001/jama.2023.18688
- Coutinho P, Martins da Silva A, Lopes Lima J, Resende Barbosa A. Forty years of experience with type I amyloid neuropathy: review of 483 cases. In: Glenner GG, Pinho e Costa P, Falcao de Freitas A, eds. *Amyloid and Amyloidosis*. *Excerpta Medica*. 1980:88-93.
- Dyck PJB, Gonzalez-Duarte A, Obici L, et al. Development of measures of polyneuropathy impairment in hATTR amyloidosis: From NIS to mNIS + 7. *J Neurol Sci*. 2019;405:116424. doi:10.1016/j.jns.2019.116424
- Elliott P, Drachman BM, Gottlieb SS, et al. Long-term survival with tafamidis in patients with transthyretin amyloid cardiomyopathy. *Circ Heart Fail*. 2022;15(1):e008193. doi:10.1161/CIRCHEARTFAILURE.120.008193
- Feng KY, Loungani RS, Rao VN, et al. Best practices for prognostic evaluation of a patient with transthyretin amyloid cardiomyopathy. *JACC CardioOncol*. 2019;1(2):273-279. doi:10.1016/j.jaccao.2019.11.006
- Fontana M, Berk JL, Gillmore JD, et al. Vutrisiran in patients with transthyretin amyloidosis with cardiomyopathy. *N Engl J Med*. 2025;392(1):33-44. doi:10.1056/NEJMoa2409134
- Galant NJ, Westermarck P, Higaki JN, Chakrabartty A. Transthyretin amyloidosis: an under-recognized neuropathy and cardiomyopathy. *Clin Sci (Lond)*. 2017;131(5):395-409. doi:10.1042/CS20160413
- Garcia-Pavia P, Bengel F, Brito D, et al. Expert consensus on the monitoring of transthyretin amyloid cardiomyopathy. *Eur J Heart Fail*. 2021;23(6):895-905. doi:10.1002/ejhf.2198
- Gertz MA, Dispenzieri A. Systemic amyloidosis recognition, prognosis, and therapy: a systematic review. *JAMA*. 2020;324(1):79-89. doi:10.1001/jama.2020.5493
- Gillmore JD, Judge DP, Cappelli F, et al. Efficacy and safety of acoramidis in transthyretin amyloid cardiomyopathy. *N Engl J Med*. 2024;390(2):132-142. doi:10.1056/NEJMoa2305434
- Hanson JLS, Avantis M, Koch CM, et al. Utility of serum transthyretin as a prognostic indicator and predictor of outcome in cardiac amyloid disease associated with wild-type transthyretin. *Circ Heart Fail*. 2019;11(2):e004000. doi:10.1161/CIRCHEARTFAILURE.117.004000



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Hellenbart EL, Ipema HJ, Rodriguez-Ziccardi MC, Krishna H, DiDomenico RJ. Disease-modifying therapies for amyloid transthyretin cardiomyopathy: Current and emerging medications. *Pharmacotherapy*. 2025;45(2):124-144. doi:10.1002/phar.4639

Institute for Clinical and Economic Review. Disease modifying therapies for the treatment of transthyretin amyloid cardiomyopathy (ATTR-CM): final evidence report. Published October 21, 2024. Accessed April 4, 2025. https://icer.org/wp-content/uploads/2024/03/ICER_ATTR-CM_Final-Report_For-Publication_10212024.pdf

Judge DP, Gillmore JD, Alexander KM, et al. Long-term efficacy and safety of acoramidis in ATTR-CM: initial report from the open-label extension of the ATTRIBUTE-CM trial. *Circulation*. 2025;151(9):601-611. doi:10.1161/CIRCULATIONAHA.124.072771

Kaku MC, Bhadola S, Berk JL, Santhorawala V, Connors LH, Lau KHV. Neurological manifestations of hereditary transthyretin amyloidosis: a focus on diagnostic delays. *Amyloid*. 2022;29(3):184-189. doi:10.1080/13506129.2022.2046557

Karam C, Mauermann ML, Gonzalez-Duarte A, et al. Diagnosis and treatment of hereditary transthyretin amyloidosis with polyneuropathy in the United States: Recommendations from a panel of experts. *Muscle Nerve*. 2024;69(3):273-287. doi:10.1002/mus.28026

Kittleson MM, Ruberg FL, Ambardekar AV, et al. 2023 ACC Expert Consensus Decision Pathway on Comprehensive Multidisciplinary Care for the Patient with Cardiac Amyloidosis: A Report of the American College of Cardiology Solution Set Oversight Committee. *J Am Coll Cardiol*. 2023;81(11):1076-1126. doi:10.1016/j.jacc.2022.11.022

Koike H, Okumura T, Murohara T, Katsuno M. Multidisciplinary approaches for transthyretin amyloidosis. *Cardiol Ther*. 2021;10(2):289-311. doi:10.1007/s40119-021-00222-w

Ladefoged B, Dybro A, Povlsen JA, Vase H, Clemmensen TS, Poulsen SH. Diagnostic delay in wild type transthyretin cardiac amyloidosis - A clinical challenge. *Int J Cardiol*. 2020;304:138-143. doi:10.1016/j.ijcard.2019.12.063

Levy JC, Teruya SL, Mirabal A, Wats K, Maurer MS, Bampatsias D. Worsening Kansas City Cardiomyopathy Questionnaire Overall Summary Score (KCCQ-os) is independently associated with increased risk of adverse outcomes in transthyretin cardiac amyloidosis (ATTR-CA). Presented at: American College of Cardiology Annual Scientific Session; March 29-31, 2025; Chicago, IL.

Martyn T, Rubio AC, Estep JD, Hanna M. Opportunities for earlier diagnosis and treatment of cardiac amyloidosis. *Methodist Debakey Cardiovasc J*. 2022;18(5):27-39. doi:10.14797/mdcvj.1163

Maurer MS, Hanna M, Grogan M, et al. Genotype and phenotype of transthyretin cardiac amyloidosis: THAOS (Transthyretin Amyloid Outcome Survey). *J Am Coll Cardiol*. 2016;68(2):161-72. doi:10.1016/j.jacc.2016.03.596

Maurer MS, Schwartz JH, Gundapaneni B, et al. Tafamidis treatment for patients with transthyretin amyloid cardiomyopathy. *N Engl J Med*. 2018;379(11):1007-1016. doi:10.1056/NEJMoa1805689



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Maurer MS, Bokhari S, Damy T, et al. Expert consensus recommendations for the suspicion and diagnosis of transthyretin cardiac amyloidosis. *Circ Heart Fail.* 2019;12(9):e006075. doi:10.1161/CIRCHEARTFAILURE.119.006075

Maurer MS, Kale P, Fontana M, et al. Patisiran treatment in patients with transthyretin cardiac amyloidosis. *N Engl J Med.* 2023;389(17):1553-1565. doi:10.1056/NEJMoa2300757

Nativi-Nicolau JN, Karam C, Khella S, Maurer MS. Screening for ATTR amyloidosis in the clinic: overlapping disorders, misdiagnosis, and multiorgan awareness. *Heart Fail Rev.* 2022;27(3):785-793. doi:10.1007/s10741-021-10080-2

Nativi-Nicolau J, Sarswat N, Fajardo J, et al. Best practices in specialized amyloidosis centers in the United States: a survey of cardiologists, nurses, patients, and patient advocates. *Clin Med Insights Cardiol.* 2021;15:11795468211015230. doi:10.1177/11795468211015230

Obi CA, Mostertz WC, Griffin JM, Judge DP. ATTR epidemiology, genetics, and prognostic factors. *Methodist Debaque Cardiovasc J.* 2022;18(2):17-26. doi:10.14797/mdcvj.1066

ONPATTRO. Prescribing information. Alnylam; 2023. Accessed March 21, 2025. https://www.accessdata.fda.gov/drugsatfda_docs/label/2023/210922s012lbl.pdf

Poli L, Labella B, Cotti Piccinelli S, et al. Hereditary transthyretin amyloidosis: a comprehensive review with a focus on peripheral neuropathy. *Front Neurol.* 2023;14:1242815. doi:10.3389/fneur.2023.1242815

Pruppers MH, Merkies IS, Faber CG, Da Silva AM, Costa V, Coelho T. The Val30Met familial amyloid polyneuropathy specific Rasch-built overall disability scale (FAP-RODS((c))). *J Peripher Nerv Syst.* 2015;20(3):319-327. doi:10.1111/jns.12120

Qarni TN, Jones FJS, Drachman B, et al. Treatment characteristics of patients with hereditary transthyretin amyloidosis: a cohort study. *Orphanet J Rare Dis.* 2024;19(1):191. doi:10.1186/s13023-024-03198-7

Rebello S, Hsu K, Nativi-Nicolau J, et al. Factors associated with financial toxicity in patients with transthyretin amyloidosis: results from Amyloidosis Research Consortium's treatment affordability patient and caregiver survey. *Amyloid.* 2025:1-10. doi:10.1080/13506129.2025.2462541

Rehman S, Masthan SS, Ibrahim R, et al. Pharmacological management of transthyretin amyloid cardiomyopathy: a scoping review. *Eur Heart J Cardiovasc Pharmacother.* 2024;10(6):547-556. doi:10.1093/ehjcvp/pvae044

Rintell D, Heath D, Braga Mendendez F, et al. Patient and family experience with transthyretin amyloid cardiomyopathy (ATTR-CM) and polyneuropathy (ATTR-PN) amyloidosis: results of two focus groups. *Orphanet J Rare Dis.* 2021;16(1):70. doi:10.1186/s13023-021-01706-7

Rozenbaum MH, Large S, Bhambri R, et al. Impact of delayed diagnosis and misdiagnosis for patients with transthyretin amyloid cardiomyopathy (ATTR-CM): a targeted literature review. *Cardiol Ther.* 2021;10(1):141-159. doi:10.1007/s40119-021-00219-5



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Schmidt HH, Waddington-Cruz M, Botteman MF, et al. Estimating the global prevalence of transthyretin familial amyloid polyneuropathy. *Muscle Nerve*. 2018;57(5):829-837. doi:10.1002/mus.26034

Sletten DM, Suarez GA, Low PA, Mandrekar J, Singer W. COMPASS 31: a refined and abbreviated composite autonomic symptom score. *Mayo Clin Proc*. 2012;87(12):1196-1201. doi:10.1016/j.mayocp.2012.10.013

Steen L, Ek B. Familial amyloidosis with polyneuropathy. A long-term follow-up of 21 patients with special reference to gastrointestinal symptoms. *Acta Med Scand*. 1983;214(5):387-397.

Stewart M, Shaffer S, Murphy B, et al. Characterizing the high disease burden of transthyretin amyloidosis for patients and caregivers. *Neurol Ther*. 2018;7(2):349-364. doi:10.1007/s40120-018-0106-z

TEGSEDI. Prescribing information. Akcea Pharmaceuticals; 2024. Accessed September 6, 2024. https://www.accessdata.fda.gov/drugsatfda_docs/label/2024/211172s014lbl.pdf

Ticau S, Sridharan GV, Tsour S, et al. Neurofilament light chain as a biomarker of hereditary transthyretin-mediated amyloidosis. *Neurology*. 2021;96(3):e412-e422. doi:10.1212/WNL.0000000000011090

Vera-Llonch M, Reddy SR, Chang E, Tarbox MH, Pollock M. The patient journey toward a diagnosis of hereditary transthyretin (ATTRv) amyloidosis. *Orphanet J Rare Dis*. 2021;16(1):25. doi:10.1186/s13023-020-01623-1

VYNDAQEL. Prescribing information. Pfizer; 2023. Accessed March 21, 2025. https://www.accessdata.fda.gov/drugsatfda_docs/label/2023/211996s002,212161s002lbl.pdf

WAINUA. Prescribing information. AstraZeneca; 2024. Accessed March 21, 2025. https://www.accessdata.fda.gov/drugsatfda_docs/label/2024/217388s002lbl.pdf

Yarlas A, Gertz MA, Dasgupta NR, et al. Burden of hereditary transthyretin amyloidosis on quality of life. *Muscle Nerve*. 2019;60(2):169-175. doi:10.1002/mus.26515