



Jaap Groothoff

Dr Groothoff is an emeritus (since April 2024) professor in paediatric nephrology with special interest in hyperoxaluria and kidney failure in children. He has been head of the department of paediatric nephrology of the Amsterdam UMC/Emma children's Hospital from 2011 up to 2023. Former positions included head of the Master Program of the Medical School of the University of Amsterdam, chair of the ESPN Registry and chair of Oxaleurope. He has been involved in hyperoxaluria research since 1994, was one of the founders of Oxaleurope and is currently still leader of the sole Dutch expertise centre for hyperoxaluria in Amsterdam and leader of the nationwide lumasiran Orphan Drug Access Protocol, which serves as a pilot study for controlled access to expensive orphan drugs. He is still involved in basic hyperoxaluria research at the Amsterdam UMC and principle investigator for all hyperoxaluria related trials.

Orchid: <https://orcid.org/0000-0002-8467-3506>. Scientific papers: 210; book chapters: 8. H-factor:42

Hyperoxaluria related papers 2020-2025

1. van Harskamp D, Garrelfs SF, Oosterveld MJS, **Groothoff JW**, van Goudoever JB, Schierbeek H. Development and Validation of a New Gas Chromatography-Tandem Mass Spectrometry Method for the Measurement of Enrichment of Glyoxylate Metabolism Analytes in Hyperoxaluria Patients Using a Stable Isotope Procedure. *Anal Chem*. 2020. PMID: 32165440
2. Milliner DS, McGregor TL, Thompson A, Dehmel B, Knight J, Roskamp R, Blank M, Yang S, Fargue S, Rumsby G, **Groothoff J**, Allain M, West M, Hollander K, Lowther WT, Lieske JC. Endpoints for Clinical Trials in Primary Hyperoxaluria. *Clin J Am Soc Nephrol*. 2020. PMID: 32165440
3. Garrelfs SF, Frishberg Y, Hulton SA, Koren MJ, O'Riordan WD, Cochat P, Deschênes G, Shasha-Lavsky H, Saland JM, Van't Hoff WG, Fuster DG, Magen D, Mochhala SH, Schalk G, Simkova E, **Groothoff JW**, Sas DJ, Meliambro KA, Lu J, Sweetser MT, Garg PP, Vaishnav AK, Gansner JM, McGregor TL, Lieske JC; ILLUMINATE-A Collaborators. Lumasiran, an RNAi Therapeutic for Primary Hyperoxaluria Type 1. *N Engl J Med*. 2021. PMID: 33830344
4. Metry EL, van Dijk LMM, Peters-Sengers H, Oosterveld MJS, **Groothoff JW**, Ploeg RJ, Stel VS, Garrelfs SF. *Pediatr Nephrol*. 2021. PMID: 33830344
5. Martin-Higuera C, Garrelfs SF, **Groothoff JW**, Jacob D, Mochhala SH, Bacchetta J, Acquaviva C, Zaniew M, Sikora P, Beck BB, Hoppe B. A report from the European Hyperoxaluria Consortium (OxalEurope) Registry on a large cohort of patients with primary hyperoxaluria type 3. *Kidney Int*. 2021. PMID: 33865885
6. Frishberg Y, Deschênes G, **Groothoff JW**, Hulton SA, Magen D, Harambat J, van't Hoff WG, Lorch U, Milliner DS, Lieske JC, Haslett P, Garg PP, Vaishnav AK, Talamudupula S, Lu J, Habtemariam BA, Erbe DV, McGregor TL, Cochat P and on behalf of the study collaborators. Phase 1/2 Study of Lumasiran for Treatment of Primary Hyperoxaluria Type 1 A Placebo-Controlled Randomized Clinical Trial. *Clin J Am Soc Nephrol*. 2021. PMID: 33985991
7. Hoppe B, Koch A, Cochat P, Garrelfs SF, Baum MA, **Groothoff JW**, Lipkin G, Coenen M, Schalk G, Amrite A, McDougall D, Barrios K, Langman CB. Safety, pharmacodynamics, and exposure-response modeling results from a first in human phase 1 study of nedosiran in primary hyperoxaluria. *Kidney Int*. 2021. PMID: 34481803
8. Garrelfs S, van Harskamp D, Peters-Sengers H, van den Akker C, Wanders R, Wijburg F, van Goudoever J, **Groothoff J**, Schierbeek H, Oosterveld M. Endogenous Oxalate Production in Primary Hyperoxaluria Type 1 Patients. *J Am Soc Nephrol* 2021. PMID: 34686543
9. Metry EL, Garrelfs SF, Peters-Sengers H, Hulton SA, Acquaviva C, Bacchetta J, Beck BB, Collard L, Deschênes G, Franssen C, Kemper MJ, Lipkin GW, Mandrile G, Mohebbi N, Mochhala SH, Oosterveld MJS, Prikhodina L, Hoppe B, Cochat P, **Groothoff JW**; OxalEurope Consortium.. Long-Term Transplantation Outcomes in Patients With Primary Hyperoxaluria Type 1 Included in the European Hyperoxaluria Consortium Registry. *Kidney Int Reports* 2022. PMID: 35155860
10. Hulton SA, **Groothoff JW**, Frishberg Y, Koren MJ, Overcash JS, Sellier-Leclerc AL, Shasha-Lavsky H, Saland JM, Hayes W, Magen D, Mochhala SH, Coenen M, Simkova E, Garrelfs SF, Sas DJ, Meliambro KA, Ngo T, Sweetser MT, Habtemariam BA, Gansner JM, McGregor TL, Lieske JC. Randomized Clinical Trial on the Long-Term Efficacy and Safety of Lumasiran in Patients With Primary Hyperoxaluria Type 1. *Kidney Int Rep*. 2021. PMID: 35257062
11. Deesker LS, Garrelfs SF, Mandrile G, Oosterveld MJS, Cochat P, Deschênes G, Harambat J, Hulton SA, Gupta A, Hoppe B, Beck BB, Collard L, Topaloglu R, Prikhodina L, Salido ED, Neuhaus TH, **Groothoff JW**, Bacchetta J. Improved outcome of infantile oxalosis over time in Europe: data from the OxalEurope Registry. *Kidney Int Rep*. 2022. PMID: 35812297
12. Mandrile G, Beck B, Acquaviva C, Rumsby G, Deesker LS, Garrelfs SF, Gupta A, Bacchetta J, **Groothoff JW**. Genetic assessment in Primary Hyperoxaluria: why it matters. *Pediatr Nephrol*. 2022. PMID: 35695965

13. Baum MA, Langman G, Cochat P, Lieske JC, Moolchala SH, Hamamoto S, Satoh H, Mourani C, Ariceta G, Torres A, Wolley M, Belostotsky V, Forbes TA, **Groothoff JW**, Hayes W, Tönshoff B, Takayama T, Rosskamp R, Russell K, Zhou J, Amrite A, Hoppe B. PHYOX2: A Pivotal Randomized Study of Nedosiran in Primary Hyperoxaluria Type 1 or 2. *Kidney Int* 2022. PMID: 36007597
14. Metry EL, Oosterveld MJS, **Groothoff JW**, Bacchetta J. The appearance of oxalate crystals in a kidney biopsy is no proof of post-transplant oxalate nephropathy in primary hyperoxaluria type 1. *Kidney Int*. 2022. PMID: 35870818
15. Michael M, **Groothoff JW**, Shasha-Lavsky H, Lieske JC, Frishberg Y, Simkova E, Sellier-Leclerc AL, Devresse A, Guebre-Egziabher F, Bakaloglu SA, Mourani C, Saqan R, Singer R, Willey R, Habtemariam B, Gansner JM, Bhan I, McGregor T, Magen D. Lumasiran for Advanced Primary Hyperoxaluria Type 1: Phase 3 ILLUMINATE-C Trial. *Am J Kidney Dis*. 2022 PMID: 35843439
16. **Groothoff JW**, Metry E, Deesker L, Garrelfs S, Acquaviva C, Almardini R, Beck BB, Boyer O, Cerkauskienė R, Ferraro PM, Groen LA, Gupta A, Knebelmann B, Mandrile G, Moolchala SS, Prytula A, Putnik J, Rumsby G, Soliman NA, Somani B, Bacchetta J. Clinical practice recommendations for primary hyperoxaluria: an expert consensus statement from ERKNet and OxalEurope. *Nature Reviews Nephrol*. 2023. PMID: 36604599 .
17. Garrelfs SF, Metry EL, van Harskamp D, Vaz FM, van den Akker CHP, Schierbeek H, **Groothoff JW**, Oosterveld MJS Glycolate oxidase inhibition by lumasiran varies between patients with primary hyperoxaluria type 1. *Kidney Int*. 2023. PMID: 36871948
18. Metry EL, Garrelfs SF, Peters-Sengers H, Vaz FM, Bijlsma JA, Neradova A, Oosterveld MJS, **Groothoff JW** Plasma oxalate and glycolate concentrations in dialysis patients with and without primary hyperoxaluria type 1. *Nephrol Dial Transplant* 2023 PMID: 36898675
19. Goldfarb DS, Lieske JC, **Groothoff J**, Schalk G, Russell K, Yu S, Vrhnjak B Nedosiran in primary hyperoxaluria subtype 3: results from a phase I, single-dose study (PHYOX4). *Urolithiasis*. 2023. PMID: 37273015
20. Elisabeth L Metry, Lisa J Deesker, Sander F Garrelfs, Michiel J S Oosterveld, Kerensa M Beekman , Marlies E A Cornelissen , Linda Koster-Kamphuis , **Jaap W Groothoff** Successful kidney-alone transplantation in a patient with PH1 on combination RNA-interference therapy. *Kidney Int*. 2023. PMID: 37349052
21. Metry EM, Garrelfs SF, Deesker LD, Acquaviva C, D'Ambrosio V, Bacchetta J, Beck BB, Cochat P, Collard L, Hogan J, Ferraro PM, Franssen CSM, Harambat J, Hulton SA, Lipkin GW, Mandrile G, Martin-Higueras C, Mohebbi N, Moolchala SH, Neuhaus TJ, Prikhodina L, Salido ED, Topaloglu R, Oosterveld MJS, **Groothoff JW**, Peters-Sengers H. Determinants of kidney failure in Primary Hyperoxaluria type 1: findings of the European Hyperoxaluria Consortium. *Kidney Int Rep*. 2023. PMID: 37849991
22. Ferraro PM, D'Ambrosio V, Gambaro G, Giachino D, **Groothoff J**, Mandrile G. A clinical screening algorithm for primary hyperoxaluria type 1 in adults on dialysis. *Nephrol Dial Transplant*. 2024. PMID: 37708050
23. Garrelfs SF, Chorny S, te Brinke H, Ruiter J, **Groothoff J**, Wanders RJA. Glyoxylate reductase: Definitive identification in human liver mitochondria, its importance for the compartment-specific detoxification of glyoxylate. *J Inherit Metab Dis*. 2024. PMID: 38200664
24. **Groothoff J**, Sellier-Leclerc AL, Deesker L, Bacchetta J, Schalk G, Tönshoff B, Lipkin G, Lemoine S, Bowman T, Zhou J, Hoppe B. Nedosiran Safety and Efficacy in PH1: Interim Analysis of PHYOX3. *Kidney Int Rep*. 2024 Mar 4;9(5):1387-1396. PMID: 38707801
25. Saland JM, Lieske JC, **Groothoff JW**, Frishberg Y, Shasha-Lavsky H, Magen D, Moolchala SH, Simkova E, Coenen M, Hayes W, Hogan J, Sellier-Leclerc AL, Willey R, Gansner JM, Hulton SA. Efficacy and Safety of Lumasiran in Patients With Primary Hyperoxaluria Type 1: Results from a Phase III Clinical Trial. *Kidney Int Rep*. 2024.PMID: 3908173
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27. Cellini B, Baum MA, Frishberg Y, **Groothoff JW**, Harris PC, Hulton SA, Knauf F, Knight J, Lieske JC, Lowther WT, Moolchala S, Nazzari L, Tasian GE, Whittamore JM, Sas DJ. Opportunities in Primary and Enteric Hyperoxaluria at the Cross-Roads Between the Clinic and Laboratory. *Kidney Int Rep*. 2024. PMID: 39534212
28. Wanders RJA, **Groothoff JW**, Deesker LJ, Salido E, Garrelfs SF. Human glyoxylate metabolism revisited: New insights pointing to multi-organ involvement with implications for siRNA-based therapies in primary hyperoxaluria. *J Inherit Metab Dis*. 2024 PMID: 39582099
29. Rosenberg N, Manders E, van den Berg S, Deesker LJ, Garrelfs SF, de Visser SJ, **Groothoff JW**, Hollak CEM. Application of four pricing models for orphan medicines: a case study for lumasiran. *Orphanet J Rare Dis*. 2024 Dec 23;19(1):485. PMID: 39716306
30. Deesker LJ, Oubram L, Almardini R, Baum MA, Bonilla-Felix M, Figueres L, Garrelfs SF, **Groothoff JW**, Ekulu PM, Müller RU, Oosterveld MJS, Qian S, Sayer JA, Soliman N, Moolchala SH, Bacchetta J. Global access to management of primary hyperoxaluria: a survey on behalf of OxalEurope, G&K working group of the ERA, and ESPN. *Nephrol Dial Transplant*. 2025. PMID: 39984743
31. Sellier-Leclerc AL, Magen D, Shasha-Lavsky H, Simkova E, Devresse A, Guebre-Egziabher F, Michael M, Lieske JC, Frishberg Y, Bakaloglu SA, Mourani C, Saqan R, Singer R, Guzzo I, Makarova N, Willey R, Kaspar C, Gansner JM, **Groothoff JW** Efficacy and Safety of Lumasiran for Advanced Primary Hyperoxaluria Type 1: 24-Month Follow-Up of the Phase 3 Illuminate-C Trial. *Am J Kidney Dis*. 2025. PMID: 40086686 .
32. Deesker LJ, Franssen CFM, Dorresteyn E, van de Kar NCAJ, Nurmohamed SA, Severs D, Garrelfs SF, Pisters-van Roy AAMG, Hollak CEM, **Groothoff JW**. Controlled access to lumasiran in primary hyperoxaluria type 1: evaluation of a new access route for orphan drugs in The Netherlands. *Nephrol Dial Transplant*. 2025. PMID: 40121182

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