



UNIKLINIK
KÖLN

Bewegungsstörungen - Was gibt es Neues?

Neuropädiatrie im Vogelflug

GNP 2023 Dortmund

Interessenskonflikt

PI in STIM-CP Studie, partielle Finanzierung durch Boston Scientific

Übersichtsartikel

Transition

Movement Disorders

CLINICAL PRACTICE

Transitional Care for Young People with Movement Disorders: Consensus-Based Recommendations from the MDS Task Force on Pediatrics

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- Zusammensetzung des interdisziplinären Team
- Zeitliche Planung
- Therapie- / Versorgungsziele
- Administration und Wissenschaftliche Aspekte

Glutaracidurie Typ 1

Received: 19 July 2022 | Revised: 28 September 2022 | Accepted: 30 September 2022

DOI: 10.1002/jimd.12566

ORIGINAL ARTICLE



Recommendations for diagnosing and managing individuals with glutaric aciduria type 1: Third revision

Nikolas Boy¹  | Chris Mühlhausen² | Esther M. Maier³ |
Diana Ballhausen⁴ | Matthias R. Baumgartner⁵ | Skadi Beblo⁶ |
Peter Burgard¹ | Kimberly A. Chapman⁷ | Dries Dobbelaere⁸ |
Jana Heringer-Seifert¹ | Sandra Fleissner³ | Karina Grohmann-Held⁹ |
Gabriele Hahn¹⁰ | Inga Harting¹¹ | Georg F. Hoffmann¹ | Frank Jochum¹² |
Daniela Karall¹³ | Vassiliki Konstantopoulous¹⁴ | Michael B. Krawinkel¹⁵ |
Martin Lindner¹⁶ | E. M. Charlotte Märtner¹ | Jean-Marc Nuoffer¹⁷ |
Jürgen G. Okun¹ | Barbara Plecko¹⁸ | Roland Posset¹ | Katja Sahn¹ |
Sabine Scholl-Bürgi¹² | Eva Thimm¹⁹ | Magdalena Walter¹ |
Monique Williams²⁰ | Stephan vom Dahl²¹ | Athanasia Ziagaki²² |
Johannes Zschocke²³ | Stefan Kölker¹

TABLE 1 Summary of all 24 recommendations

	Level of recommendation ^a
Diagnostisches Vorgehen	Strong recommendation for (A)
2 Positive (abnormal) NBS results and/or suggestive clinical, biochemical and/or neuroradiological signs should be confirmed by diagnostic work-up, including quantitative analysis of GA and 3-OH-GA in urine and/or blood, and, if abnormal, molecular genetic analysis of <i>GCDH</i> gene and/or GCDH enzyme analysis in leukocytes or fibroblasts (Figure 1).	Strong recommendation for (A)
3 In children with SDH/hydronephrosis (fluid collections) in combination with further characteristic neuroradiologic signs (frontotemporal hypoplasia with widening of anterior temporal CSF spaces and the Sylvian fissure, Table S2), targeted diagnostic work-up (using the algorithm in Figure 1) is strongly recommended.	Strong recommendation for (A)
4 In children with a positive (abnormal) NBS result, but negative (normal) confirmatory diagnostic work-up, the mother may be informed about the possible condition of a maternal GA1 which can be further examined by targeted diagnostic work-up (Figure 1).	Recommendation for research (O)
5 Patients should be regularly evaluated by an expert centre in managing inherited metabolic diseases.	Strong recommendation for (A)
6 Up to the age of 6 years. To ensure sufficient protein intake, additional administration of lysine-free, tryptophan-reduced and arginine-enriched amino acid mixtures is strongly recommended.	Strong recommendation for (A)
7 After age 6 years, dietary treatment should follow an age-adapted, protein-controlled protocol which is based on safe levels for protein intake and avoids excessive intake of food with high lysine content. Dietary transition should be accompanied by regular dietary advice.	Recommendation for (B)
8 Since there is no evidence for clinical benefit of the use of arginine as a single amino acid for maintenance or emergency treatment in addition to arginine intake via natural food and AAM, an additional arginine supplementation is not recommended.	Recommendation for research (O)
9 Carnitine should be supplemented lifelong aiming to maintain the concentration of free carnitine in the normal range.	Recommendation for (B)
10 Patients should be treated immediately and to perform it aggressively in any case of febrile illness, or alarming symptoms as well as during perioperative management within the vulnerable period for striatal injury (up to age 6 years).	Strong recommendation for (A)
11 Emergency treatment after age 6 years can be administered during episodes of severe illness or perioperative management in analogy to the age group 0–6 years with individual adaptation of treatment.	Recommendation for research (O)
12 Patients with frequent (more than 2 times per year) tonic-clonic epileptic seizures or neurosurgically treatable manifestations (SDH) should be managed by a neuropaediatrician/neurologist and/or neurosurgeon in close cooperation with metabolic specialists.	Strong recommendation for (A)
13 Patients should be vaccinated according to national recommendations.	Recommendation for (B)
14 Patients and their families on disease course, treatment and prognosis as well as socio-legal advice and evaluation of quality of life should be regularly provided by an interdisciplinary team including experts in metabolic medicine, nutritional therapy, physiotherapy, social-advice and psychology.	Recommendation for (B)

Klinische Überwachung

15 Therapeutic effectiveness and adverse side effects should be monitored by regular follow-up investigations and intensified in case of symptom progress or non-adherence to treatment recommendations. For recommended endpoints of clinical monitoring see recommendations #17–20, 23, 24 and Table 6.	Strong recommendation for (A)
16 Analysis of urinary concentrations of GA and 3-OH-GA should not be used for monitoring or adaption of treatment.	Recommendation for (B)
17 Concentrations of plasma amino acids should be regularly quantified in patients with low lysine diet (3–4 h postprandially) and be maintained within the age-specific normal range (Table 5).	Strong recommendation for (A)
18 Concentration of free carnitine in plasma or dried blood spots should be monitored regularly in all individuals with GA1. Trough level concentration of free carnitine (at least 12 h after last administration) should be maintained within the reference range.	Recommendation for (B)
19 Renal function should be assessed yearly starting from age 6 years (Table 7).	Recommendation for (B)
20 Patients should be admitted to a hospital and closely monitored for at least 24 h even after minimal or mild head trauma within the first 3 years of life due to the increased risk for developing SDH.	Recommendation for (B)
21 Neuroradiological examination should be performed in all age groups if neurological symptoms occur or deteriorate significantly.	Recommendation for (B)
22 Routine MRI investigations for detection and/or monitoring of extrastriatal abnormalities (subependymal noduli, white matter abnormalities) can be started from age 10 years and repeated depending on results, for example, every 2–5 years).	Recommendation for research (O)
23 Intelligence/developmental quotient, motor functions and language should be evaluated regularly to detect specific deficits and allow start of supportive treatment. For severely affected patients adjusted (Table 6).	Recommendation for (B)

Transition

24 Depending on local health care structures, transition (interdisciplinary paediatric-internal consultation) followed by transfer to adult medicine should be broached and organised as a structured and standardised procedure.	Recommendation for (B)
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^aLevel of recommendation according to.^{45,66}

AAV2-Gentherapie bei AADC-Mangel

Received: 26 April 2023 | Revised: 29 June 2023 | Accepted: 3 July 2023
DOI: 10.1002/jimd.12649

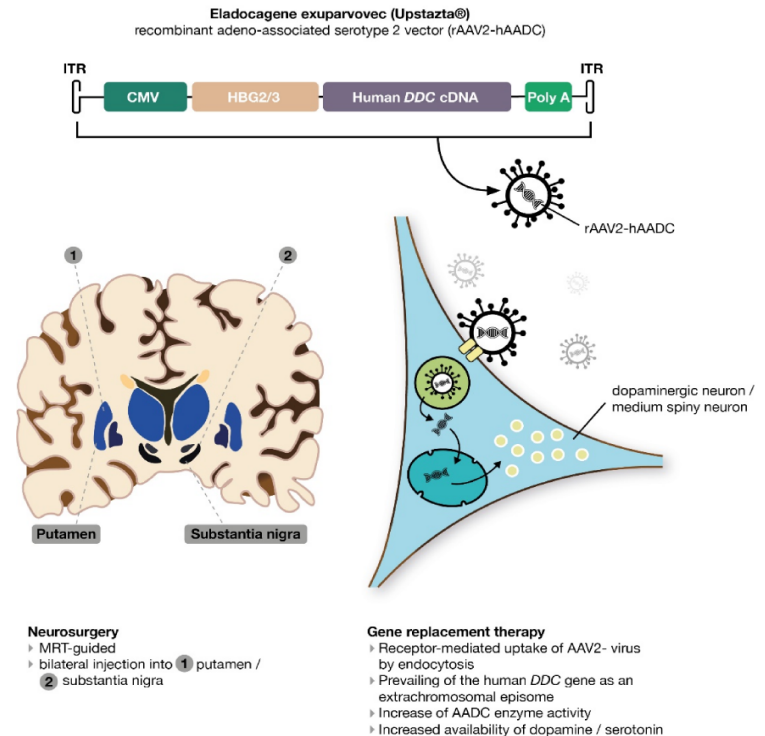
ORIGINAL ARTICLE

JIMD | WILEY

Gene therapy for aromatic L-amino acid decarboxylase deficiency: Requirements for safe application and knowledge-generating follow-up

Agathe Roubertie¹ | Thomas Opladen² | Heiko Brennenstuhl^{2,3} |
Oya Kuseyri Hübschmann² | Lisa Flint⁴ | Michel A. Willemsen⁵ |
Vincenzo Leuzzi⁶ | Angels Garcia Cazorla⁷ | Manju A. Kurian^{8,9} |
Marie Céline François-Heude¹ | Paul Hwu¹⁰ | Bruria Ben Zeev^{11,12} |
Karl Kiening¹³ | Thomas Roujeau¹⁴ | Roser Pons¹⁵ | Toni S. Pearson¹⁶

- Gentherapie mit Eladocogene exuparvovec EMA zugelassen
- International Working Group on Neurotransmitter Related Disorders (iNTD)
- Standardisierte Empfehlungen zur Vorbereitung, Behandlung und Follow-up der Patienten



Cerebralpalse

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August 09, 2022; 99 (6) CONTEMPORARY ISSUES IN PRACTICE, EDUCATION, & RESEARCH

Top 10 Research Themes for Dystonia in Cerebral Palsy

A Community-Driven Research Agenda

Laura A. Gilbert, Darcy L. Fehlings, Paul Gross, Michael C. Kruer, Wendy Kwan, Jonathan W. Mink, Michele Shusterman, Bhooma R. Aravamuthan, and the Cerebral Palsy Research Network Dystonia Study Group

Institution: DEUTSCHE ZENTRUM FÜR NEUROLOGIE

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Table 2 Top 10 Research Themes for DCP

Theme	Uncertainties	Rank	Score (%)	Source
1. Develop new treatments for individuals with DCP	Can we find more effective medications for dystonia with fewer side effects and longer therapeutic effects than currently available treatments?	1	69.68	Community member
	How can we better improve function and outcomes for those with dystonia as part of their CP?	5	64.04	Clinician
	Can stem cell treatment improve outcomes for dystonic CP?	26	50.68	Community member
2. Assess rehabilitation, psychological, and environmental approaches to manage dystonia	What are the nonmedical interventions that can help reduce dystonia?	2	66.46	Community member
	What are the most effective treatments for dystonia including pharmacologic, surgical, and nonpharmacologic therapies?	3	64.44	Researcher
3. Compare effectiveness of pharmacologic and surgical treatments for dystonia (including evaluation of side effects, a person's overall function, and effect on individualized goals)	What are the long-term health effects and outcomes of treatments used for dystonia (including intrathecal badofen) and DBS?	4	64.06	Clinician
	Do first-line dystonia medications interfere with learning and cognitive performance?	8	58.33	Community member
	What is the most effective way to use rehabilitation strategies as an adjunct for medical or surgical treatments?	16	54.77	Clinician
	Is DBS or ITB more effective in treating DCP?	19	52.97	Clinician
	What is the role of medical marijuana and cannabidiol in patients with dystonia?	21	52.13	Clinician
	What are the adverse effects of medications and how do we assess for these in patients who are unable to effectively communicate?	25	51.33	Clinician
4. Improve the clinical consistency of dystonia diagnosis and severity assessments	Can a dystonia severity scale be made to be reliable for DCP and feasible to implement in clinic?	6	60.35	Community member
	How can we increase the consistency of care from clinicians seeing children with DCP?	12	56.14	Community member
	Why is dystonia inconsistently diagnosed across providers?	22	51.91	Clinician
5. Assess the effect of mixed tone (spasticity and dystonia) in CP in outcomes and approaches	In patients with mixed spasticity and dystonia, can we determine the treatable elements of spasticity and dystonia that lead to hip and spine deformities?	7	58.50	Community member
6. Assess predictors of treatment responsiveness (e.g. etiology, severity, earlier detection) in individuals with DCP	Does dystonia etiology or severity play a role in treatment efficacy?	9	57.88	Community member
	Does earlier detection and intervention in dystonia lead to improved outcomes?	10	57.11	Community member
	How does dystonia of varying severity affect recovery from surgical or medical interventions?	29	50.14	Community member
7. Identify what causes DCP (the pathophysiologic mechanism)	What is the pathophysiologic mechanism of DCP?	13	56.09	Researcher
	Is dystonia related to unorganized motor patterns driven by plasticity?	27	50.26	Clinician
	What percentage of patients with dystonic CP have an underlying genetic etiology?	28	50.22	Researcher
8. Characterize the natural history of DCP	What is the natural history of patients with nonprogressive dystonia?	14	56.02	Community member
9. Determine the best treatments for pain due to dystonia in CP	What are the best treatments for pain secondary to dystonia?	15	55.28	Clinician
10. Increase awareness of DCP among families	Is there a need for a unified dystonia guide for parents and medical professionals? (e.g., the CP Toolkit)	23	51.51	Community member
	What education can we provide to physicians and families to improve the awareness and diagnosis of dystonia?	24	51.50	Community member

Abbreviations: CP = cerebral palsy; DBS = deep brain stimulation; DCP = dystonia in CP; ITB = intrathecal badofen.

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Klinische Studien

Funktionelle Tic-Störung

European journal of neurology
the official journal of the European Academy of Neurology



ORIGINAL ARTICLE | [Full Access](#)

European Society for the Study of Tourette Syndrome 2022 criteria for clinical diagnosis of functional tic-like behaviours: International consensus from experts in tic disorders

Tamara Pringsheim ✉, Christos Ganos, Christelle Nilles, Andrea E. Cavanna, Donald L. Gilbert, Erica Greenberg, Andreas Hartmann, Tammy Hedderly, Isobel Heyman, Holan Liang ... [See all authors](#) ▾

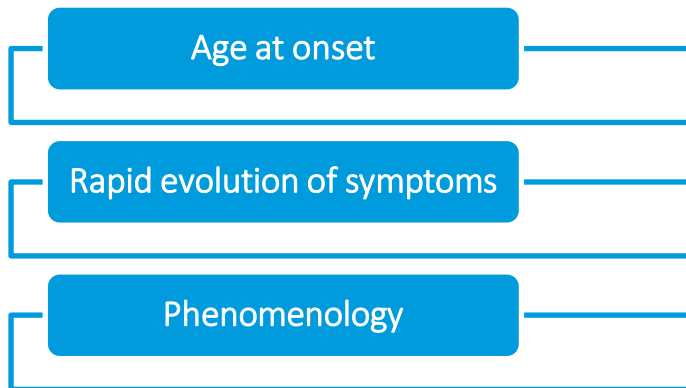
First published: 01 January 2023 | <https://doi.org/10.1111/ene.15672> | Citations: 8

Hintergrund:

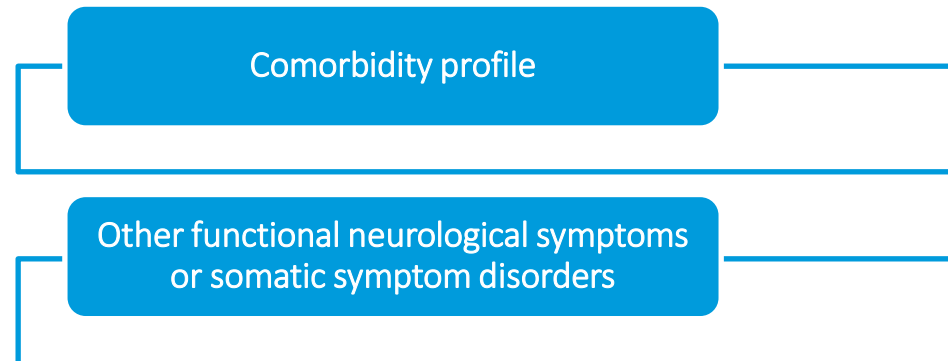
- Deutlicher Anstieg funkt. Tic-Störungen 2020
- 24 Experten, web-basierte Delphi-Survey

- **2 Major + 1 Minor Criteria: V.a. funkt.Tic-Störung**
- **Alle 3 Major Criteria: Diagnose funkt. Tic-Störung**

Major criteria



Minor criteria



Hyperkinetische Bewegungsstörungen



Research Article | [Open Access](#) |

The Genetic Landscape of Complex Childhood-Onset Hyperkinetic Movement Disorders

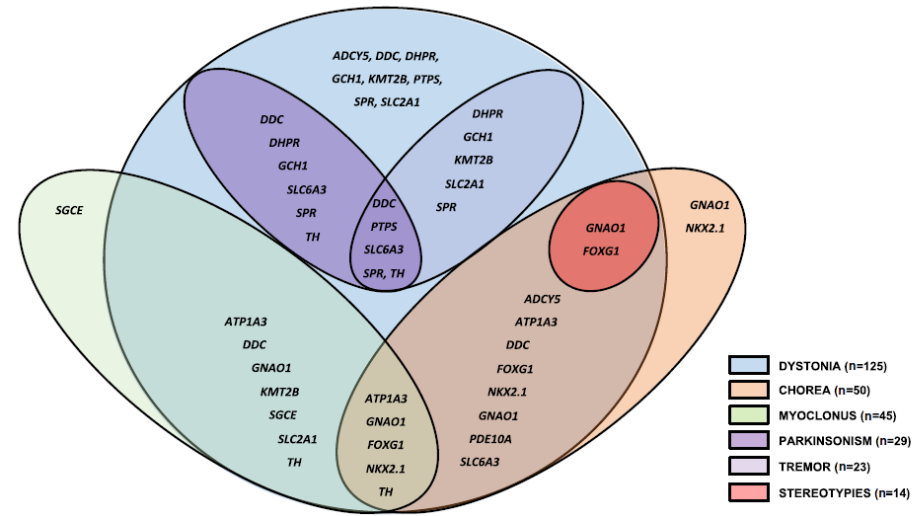
Belén Pérez-Dueñas MD, PhD Kathleen Gorman MD, Anna Marcé-Grau PhD, Juan D. Ortigoza-Escobar MD, PhD, Alfons Macaya MD, PhD, Federica R. Danti MD ... [See all authors](#)

Studiendesign:

- Retrospektive Datenanalyse
- Multizentrisch
- 140 Pat. mit komplex. Bewegungsstörung
- mittl. Alter bei Erfassung 13,3 Jahre (0,5-68)

TABLE 2 Prevalence of different movement phenotypes in complex genetic hyperkinetic movement disorders

Genes	Chorea	Dystonia	Myoclonus	Tremor	Parkinsonism	Ataxia	Stereotypies
ADCY5							
ATP1A3							
FOXP1							
GNAO1							
KMT2B							
MICU1							
NKX2.1							
PDE10A							
SGCE							
SLC2A1							
Neurotransmitter defects							
DDC							
DHPR							
GCH							
PTPS							
SLC6A3							
SPR							
TH							
0-24%		25-49%		50-74%		75-100%	







Hyperkinetische Bewegungsstörungen - GNAO1

Movement disorders

Original research


Genotype–phenotype correlation and treatment effects in young patients with *GNAO1*-associated disorders

Moritz Thiel ,¹ Daniel Bamborschke,² Wibke G. Janzarik,³ Birgit Assmann,⁴ Simone Zittel ,⁵ Steffi Patzer,⁶ Andrea Auhuber,⁷ Joachim Opp ,⁸ Eva Matzker,⁹ Andrea Bevot,¹⁰ Juergen Seeger ,¹¹ Andreas van Baalen,¹² Burkhard Stüve,¹³ Knut Brockmann,¹⁴ Sebahattin Cirak,¹⁵ Anne Koy¹⁶


Movement Disorders

Official Journal of the International Parkinson and Movement Disorder Society



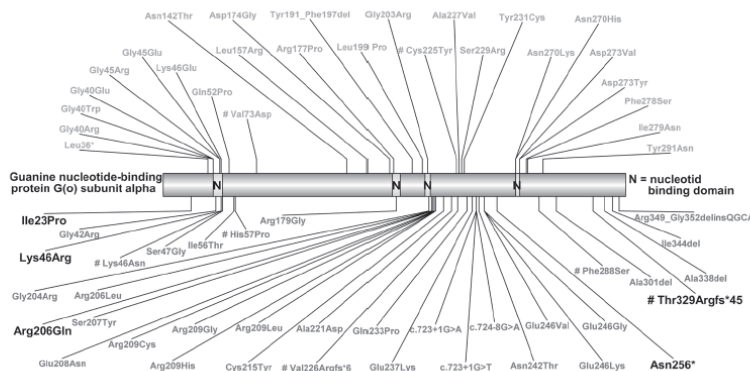
Brief Report |  Open Access |  

Highlighting the Dystonic Phenotype Related to *GNAO1*

Thomas Wirth MD , Giacomo Garone MD, Manju A. Kurian PhD, Amélie Piton PhD, Francisca Millan MD, Aida Telegrafi MS, Nathalie Drouot Msc, Gabrielle Rudolf PhD, Jamel Chelly MD ... [See all authors](#) ▾

First published: 20 June 2022 | <https://doi.org/10.1002/mds.29074> | Citations: 11

Developmental and epileptic encephalopathy phenotype



N=25 Patienten mit mildem Phänotyp:

- Späte Manifestation (>2 Jahre)
- Keine epilept. Enzephalopathie
- Freies Gehen
- Alter 23,8 Jahre (5-66 Jahre)

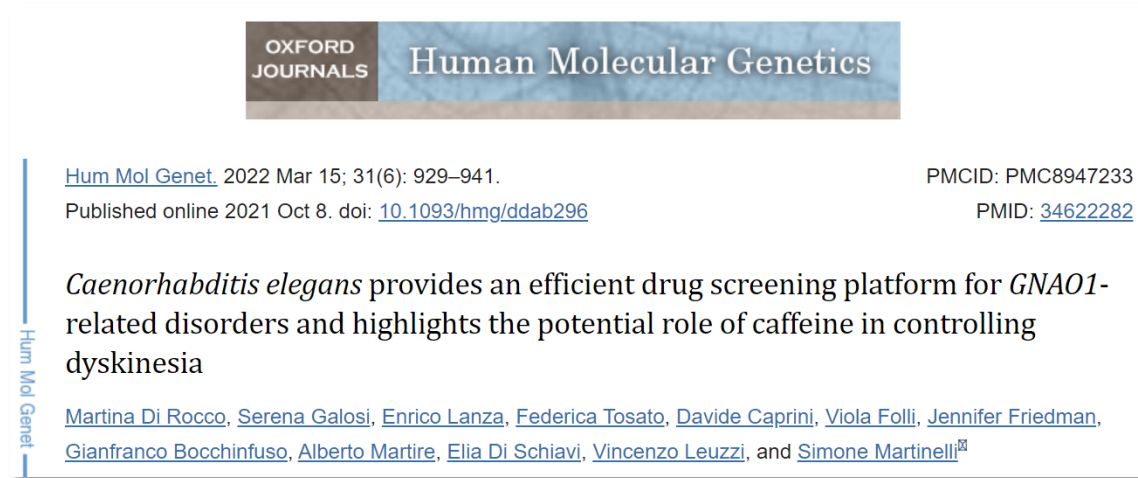
Studiendesign:

- ESNEK-basierte Umfrage
- Retrospektive, multizentrische Kohorte, n=25
- Alter 9 Jahre (4 Mo-24 Jahre)

- Erweiterung des klinischen Phänotyps
- Keine eindeutige Genotyp-Phänotyp Korrelation
- Tiefe Hirnstimulation effektiv



Hyperkinetische Bewegungsstörungen - GNAO1



- Koffein wirkt antagonistisch am Adenosin-Rezeptoren
- Funktionelle Analyse c.139A>G und c.662C>A im *C. elegans* Model

Deutliche Besserung der Bewegungsstörung durch Koffein

Hyperkinetische Bewegungsstörungen – ADCY5



Brief Report | Full Access

Efficacy of Caffeine in ADCY5-Related Dyskinesia: A Retrospective Study

Aurélie Méneret MD, PhD , Shekeeb S. Mohammad PhD, Laura Cif MD, PhD, Diane Doummar MD, Claudio DeGusmao MD, Mathieu Anheim MD, PhD, Magalie Barth MD ... [See all authors](#) ▾

First published: 05 April 2022 | <https://doi.org/10.1002/mds.29006> | Citations: 11



Molecular Genetics and Metabolism

Volume 138, Issue 1, January 2023, 106970



Deep brain stimulation effect in genetic dyskinetic cerebral palsy: The case of ADCY5- related disease

[Laura Cif](#)^a , [Diane Demailly](#)^a, [Claire Gehin](#)^a, [Emilie Chan Seng](#)^a, [Morgan Dornadic](#)^{a b}, [Sophie Huby](#)^{a b}, [Gaetan Poulen](#)^a, [Agathe Roubertie](#)^c, [Matthieu Villessot](#)^{a b}, [Thomas Roujeau](#)^a, [Philippe Coubes](#)^a

Studiendesign:

- Retrospektive Kohortenstudie
- N=30 Patienten, Alter 4-76 Jahre
- **Prim. Zielpunkt Verbesserung der Bewegungsstörung >40% (Frequenz, Dauer)**
- Dosis 60-800mg/Tag in 1-4 ED

- Koffein ist gut verträglich und effektiv
- THS kann Bewegungsstörung verbessern
- THS reduziert hyperkinetische Exazerbationen

Studiendesign:

- Retrospektive Fallserie
- 4 Patienten, Alter 13 Jahre +/- 2,9 Jahre
- Alter bei THS-GPi 9 Jahre

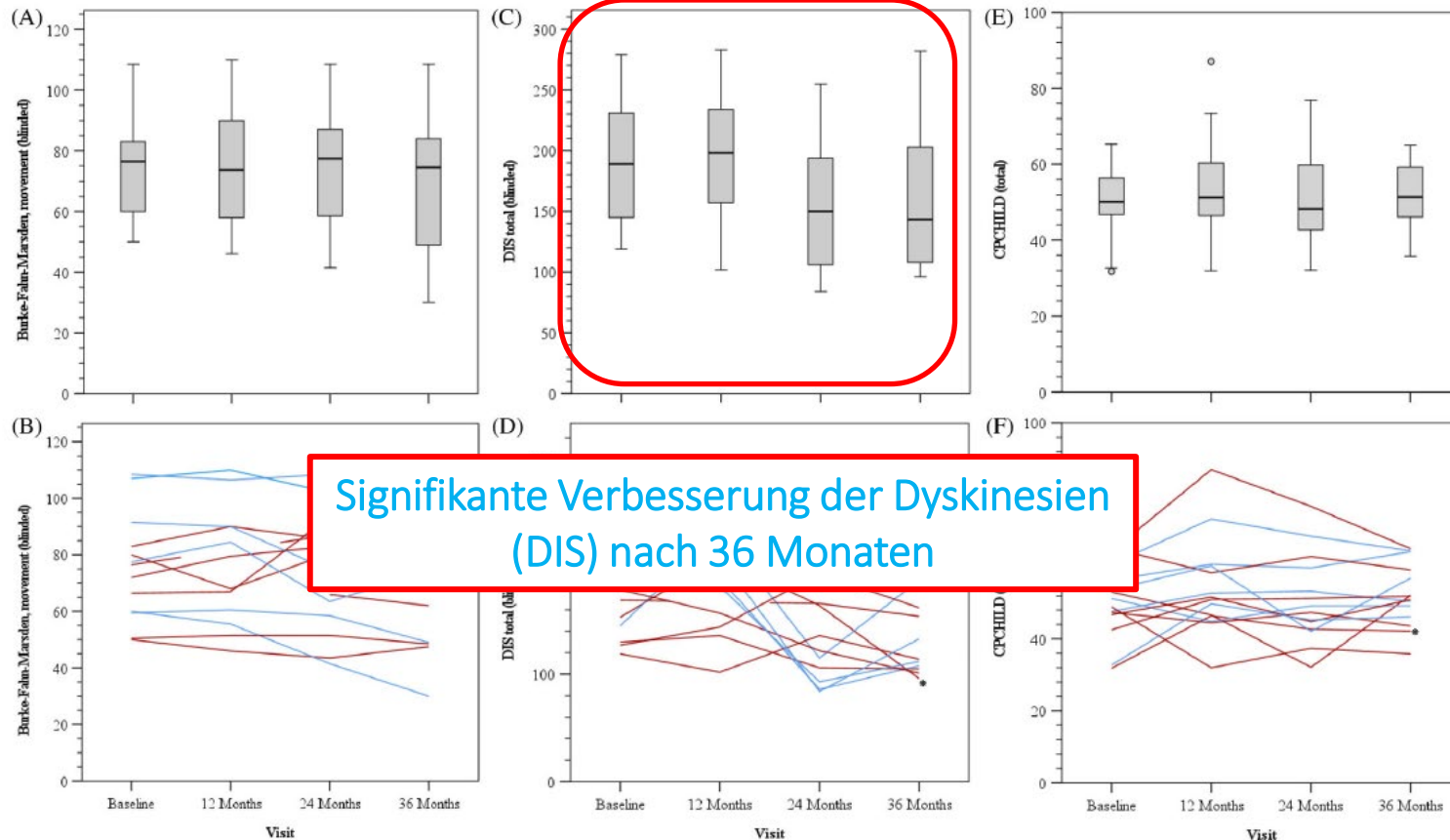
Dyskinetische CP und Tiefe Hirnstimulation



Long-Term Follow-Up of Pediatric Patients with Dyskinetic Cerebral Palsy

Anne
Matth

First p



Studiendesign

- Prospektive multizentrische Studie
- 16 Patienten mit dysk. CP
- THS-GPi
- Alter 7-18 Jahre


Alternierende Hemiplegie

Movement Disorders

Official Journal of the International
Parkinson and Movement Disorder Society

LETTERS: NEW OBSERVATIONS | [Open Access](#) | 

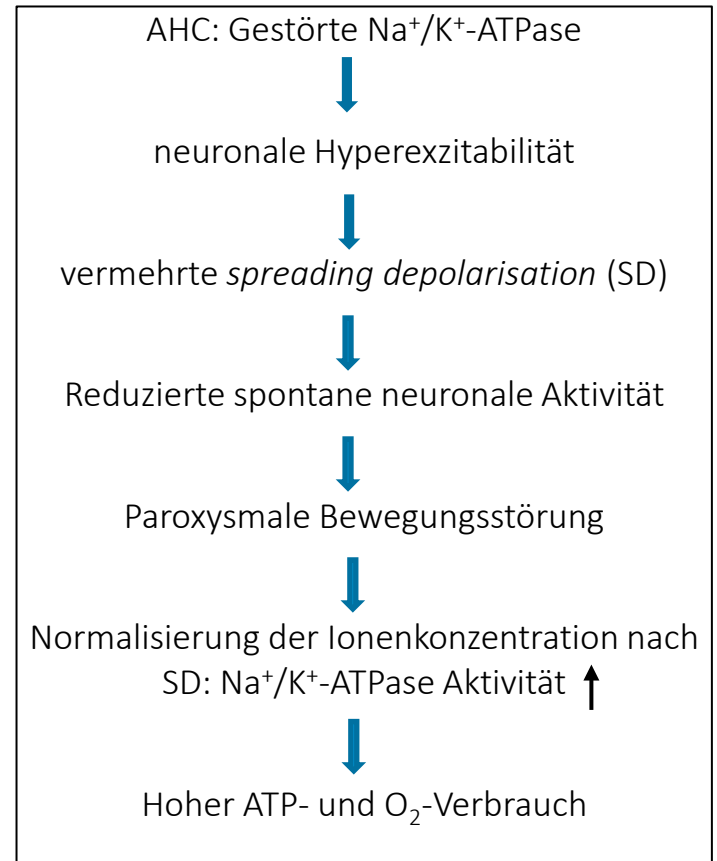
Oxygen Therapy: An Acute Treatment for Paroxysmal Dystonia in Alternating Hemiplegia of Childhood?

Quentin Welniarz PhD, Domitille Gras MD, Agathe Roubertie MD, PhD, Maria T. Papadopoulou MD, Eleni Panagiotakaki MD, PhD, Emmanuel Roze MD, PhD 

First published: 16 February 2023 | <https://doi.org/10.1002/mds.29357> | Citations: 1

Studiendesign:

- Klinische Fallserie
- N=2 Patienten, Alter 12 und 9 Jahre
- High Flow 12 l 100% O₂ über 15 Min.
- Anwendung in 60 Episoden



Verbesserung > 90% der Attacken nach 20 Minuten

Tic / Tourette-Syndrom

Volume 151, Issue 2
February 2023

ARTICLES | JANUARY 11 2023

Ecopipam for Tourette Syndrome: A Randomized Trial

Donald L. Gilbert, MD, MS ; Jordan S. Dubow, MD; Timothy M. Cunniff, PharmD; Stephen P. Wanaski, PhD; Sarah D. Atkinson, MD; Atul R. Mahabeshwarkar, MD

Address correspondence to Donald L. Gilbert, MD, MS, Division of Neurology, ML #2015 Cincinnati Children's Hospital Medical Center, 3333 Burnet Ave, Cincinnati, OH 45229. E-mail:


donald.gilbert@cchmc.org

Pediatrics (2023) 151 (2): e2022059574.

<https://doi.org/10.1542/peds.2022-059574> **Article history** 



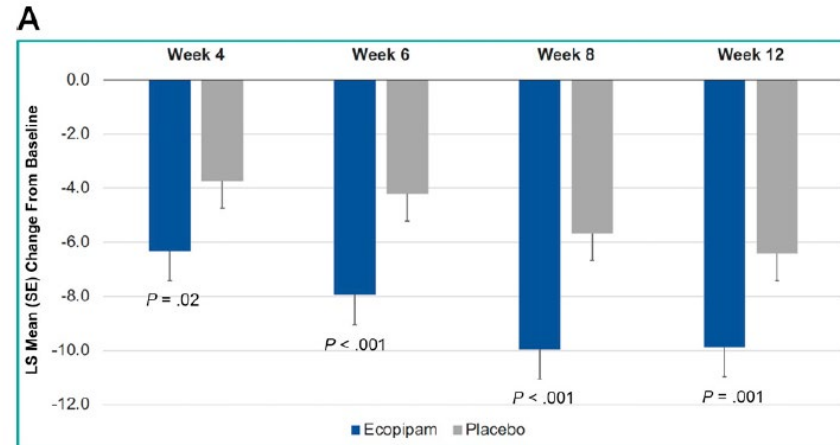
Ecopipam:

- Selektiver D1-Rezeptor-Antagonist
-  Dopaminansprechen der Neurone

Studiendesign:

- Multizentrent., doppel-verbl. Plazebo-kontrollierte Studie
- Studiendauer 12 Wochen
- N=76 versus 77 Patienten, Alter 6 – 17 Jahre
- Prim. Zielparmeter: YGTSS

- Sign. Verbesserung der motorischen und vokalen Tics
- Sicherheitsprofil gut



Tic / Tourette-Syndrom

Psychiatry Research 323 (2023) 115135

Contents lists available at ScienceDirect

Psychiatry Research

journal homepage: www.elsevier.com/locate/psychres

CANNA-TICS: Efficacy and safety of oral treatment with nabiximols in adults with chronic tic disorders – Results of a prospective, multicenter, randomized, double-blind, placebo controlled, phase IIIb superiority study

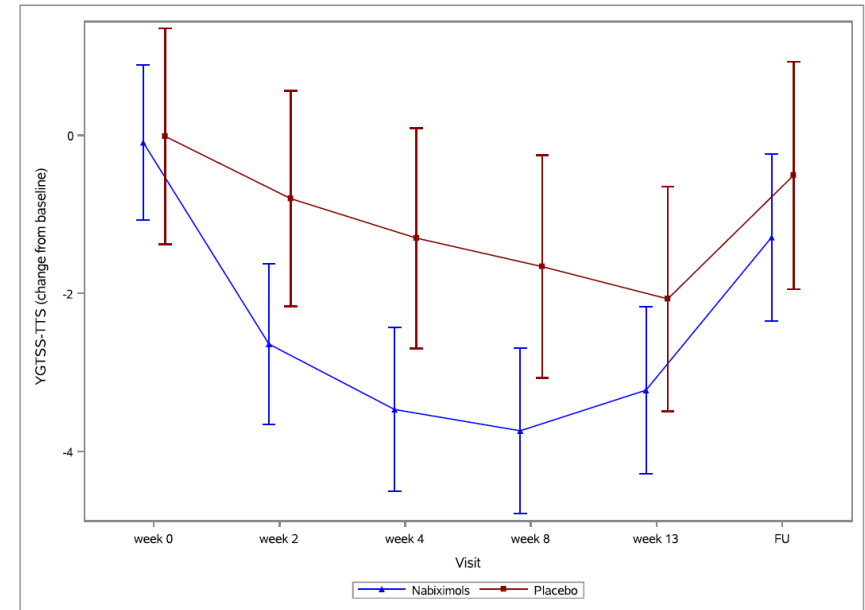
Kirsten R. Müller-Vahl^a, Anna Pisarenko^a, Natalia Szejko^{b,c}, Martina Haas^a, Carolin Fremer^a, Ewgeni Jakubovski^a, Richard Musil^d, Alexander Münchau^e, Irene Neuner^{f,g,h}, Daniel Huys^{i,j}, Ludger Tebartz van Elst^k, Christoph Schröder^l, Rieke Ringlstetter^m, Armin Koch^m, Eva Beate Jenz^m, Anika Großhennig^m



- Nabiximol: Verbesserung 21.9% versus 9.1%, n.s.
- Überlegenheit bezüglich sek. Outcome-Parameter

Studiendesign:

- Multizentrische, doppel-verblindete Plazebo-kontrollierte Phase IIIb Studie
- Adulte Patienten mit chron Tic-Störung (n=97)
- Studiendauer 13 Wochen
- **Prim. Zielparameter: YGTSS**



Friedreich Ataxie



Research Article | [Open Access](#) |

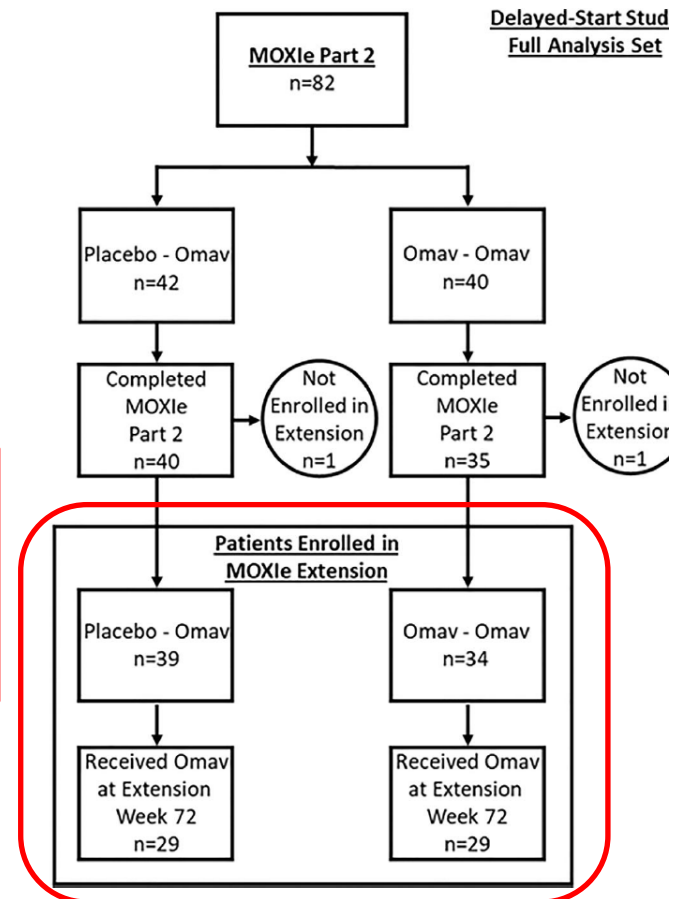
Efficacy of Omaveloxolone in Friedreich's Ataxia: Delayed-Start Analysis of the MOXle Extension

David R. Lynch MD, PhD , Melanie P. Chin PhD, Sylvia Boesch MD, Martin B. Delatycki MD, PhD, Paola Giunti MD, PhD, Angie Goldsberry MS, J. Chad Hoyle MD, Caterina Mariotti MD ... [See all authors](#) ▾

First published: 29 November 2022 | <https://doi.org/10.1002/mds.29286> | Citations: 7

- Omaveloxolone verhindert Krankheitsverschlechterung bei FA im Langzeit-Follow-up
- Früher Therapiebeginn sinnvoll
- Verträglichkeit gut

- Open-label extension study von MOXle
- N=73 Patienten, Alter 16-40 Jahre



Chorea Huntington



THE LANCET Neurology



Volume 22, Issue 6, June 2023, Pages 494-504

Articles

Safety and efficacy of valbenazine for the treatment of chorea associated with Huntington's disease (KINECT-HD): a phase 3, randomised, double-blind, placebo-controlled trial

Prof Erin Furr Stimming MD^a  , Daniel O Claassen MD^b, Elise Kayson MS^c, Jody Goldstein BS^c, Raja Mehanna MD^a, Hui Zhang PhD^d, Grace S Liang MD^d, Dietrich Haubenberger MD^d

Huntington Study Group KINECT-HD Collaborators*

- Valbenazine: Hochselektiver vesikulärer Monoamintransporter2 Hemmer (VMAT2)
- Phase 3, **random.**, **doppelblinde**, Plazebo-kontrollierte Studie (USA, Canada)
- N= 125 Patienten (adult)
- **Prim. Zielpunkt:** Verbesserung Unified Huntington's Disease Rating Scale [UHDRS] total Maximal Chorea [TMC]
- Baseline - 12 Wochen

Valbenazine führte zu sign. Verbesserung der Chorea bei Patienten mit Chorea Huntington

Genetik



Letters: New Observations | [Open Access](#) | [CC](#) | [i](#)

Both Heterozygous Variants Are Associated with Early-Onset Dystonia

Dora Steel BMBCh, Aika Frances M. Gibbon MB, ...



Brief Communication | [Open Access](#) | [CC](#) | [i](#) | [e](#) | [s](#)

Recessive *NUP54* Variants Underlie Early-Onset Dystonia with Striatal Lesions

Journal of Neurology (2023) 270:2197–2203
<https://doi.org/10.1007/s00415-023-11564-x>

ORIGINAL COMMUNICATION



Rare variants in *IMPDH2* cause autosomal dominant dystonia in Chinese population

Junyu Lin¹ · Chunyu Li¹ · Yiyuan Cui¹ · Yanbing Hou¹ · Lingyu Zhang¹ · Ruwei Ou¹ · Qianqian Wei¹ · Kuncheng Liu¹ · Tianmi Yang¹ · Yi Xiao¹ · Qirui Jiang¹ · Bi Zhao¹ · Jing Yang¹ · Xueping Chen¹ · Huifang Shang¹



Letters: New Observation | [Open Access](#) | [CC](#) | [i](#)

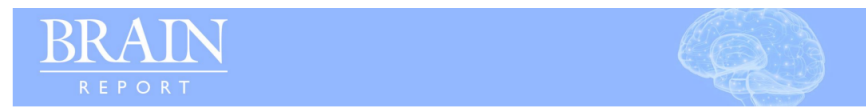
MDSGene: Extending the List of Isolated Dystonia Genes by *VPS16*, *EIF2AK1*

<https://doi.org/10.1093/brain/awad068>

BRAIN 2023; 146; 2730–2738 | 2730

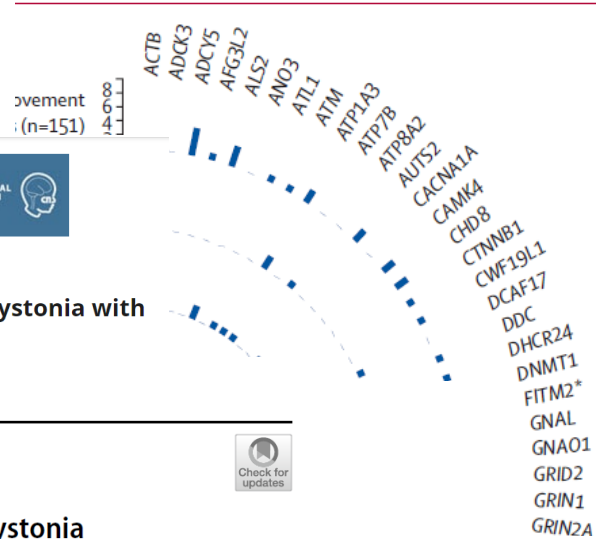
Mirja Thomsen MSc,

First published: 20 January 2023



Variants in *ATP5F1B* are associated with dominantly inherited dystonia

Alessia Nasca,^{1,†} Nicolò E. Mencacci,^{2,†} Federica Invernizzi,¹ Michael Zech,^{3,4} Ignacio J. Keller Sarmiento,² Andrea Legati,¹ Chiara Frascarelli,¹ Bernabe I. Bustos,² Luigi M. Romito,⁵ Dimitri Krainc,² Juliane Winkelmann,^{3,4,6,7} Miryam Carecchio,^{1,8,9} Nardo Nardocci,⁹ Giovanna Zorzi,⁹ Holger Prokisch,^{3,4,†} Steven J. Lubbe,^{2,†} Barbara Garavaglia,^{1,†} and Daniele Ghezzi^{1,10,†}



TUBB4B
TOR1A
THAP1



RESEARCH ARTICLE | [Open Access](#) | [CC](#) | [i](#) | [e](#) | [s](#)

Dystonia Linked to *EIF4A2* Haploinsufficiency: A Disorder of Protein Translation Dysfunction

Philip Harrer MD, Matej Škorvánek MD, PhD, Volker Kittke MSc, Ivana Dzinovic MSc, Friederike Borngräber MD, Mirja Thomsen MSc, Vanessa Mandel MSc ... See all authors

First published: 23 July 2023 | <https://doi.org/10.1002/mds.29562>



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Vielen Dank!

Klinische Studien – COVID19 Tics/Tic-ähnliche Störungen



Ergebnisse:

- 94% weiblich
- Mittleres Alter 13,7 Jahre
- Psychiatrische und entwicklungsneurologische Auffälligkeiten in 91%, Angststörung bei 67%

- Retrospektive, monozentrische Studie
- N=34 Patienten mit plötzlicher Tic-Manifestation

Starke Prävalenz plötzlicher Tic-Störungen bei überwiegend weiblichen Teenagern während der Pandemie

Übersichtsartikel – ASMs und Bewegungsstörungen

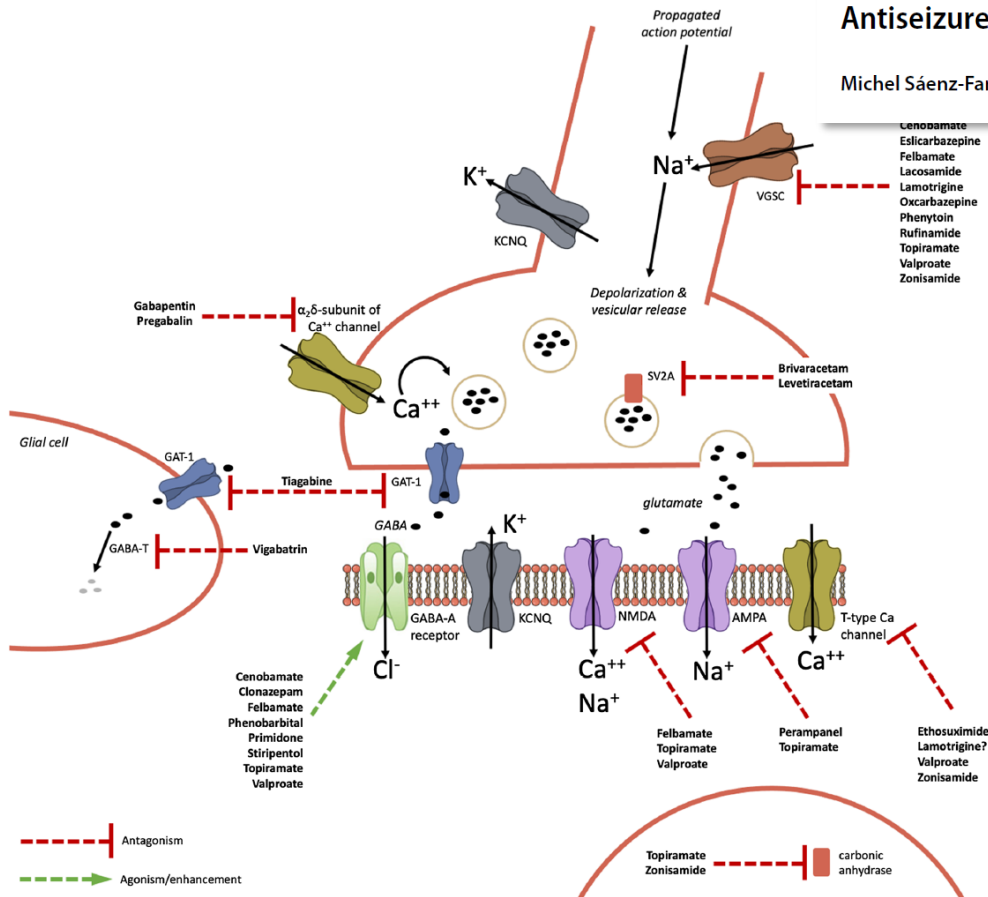
CNS Drugs (2022) 36:859–876
<https://doi.org/10.1007/s40263-022-00937-x>

REVIEW ARTICLE



Antiseizure Drugs and Movement Disorders

Michel Sáenz-Farret¹ · Marina A. J. Tijssen^{2,3} · Dawn Eliashiv⁴ · Robert S. Fisher⁵ · Kapil Sethi⁶ · Alfonso Fasano^{1,7,8}



Key Points

The relationship between antiseizure drugs and movement disorders is complex.

Antiseizure drugs are typically used for tremor, myoclonus, and restless leg syndrome.

Antiseizure drugs also represent a potential cause of iatrogenic movement disorders (parkinsonism and tremor mainly).

Antiseizure drugs with a variable effect on movement disorders are carbamazepine and valproate.

No effect on movement disorders has been reported for brivaracetam, eslicarbazepine, lacosamide, and stiripentol.

Klinische Studien – Ataxia Teleangiectasia



Official Journal of the International
Parkinson and Movement Disorder Society

Brief Report | [Open Access](#) | CC BY-NC-ND

Nicotinamide Riboside Improves Ataxia Scores and Immunoglobulin Levels in Ataxia Telangiectasia

Stefanie J.G. Veenhuis MD , Nienke J.H. van Os MD, PhD, Anjo J.W.M. Janssen PhD, Marjo H.J.C. van Gerven MSc, Karlien L.M. Coene PhD, Udo. F.H. Engelke PhD ... [See all authors](#) ▾

First published: 13 September 2021 | <https://doi.org/10.1002/mds.28788> | Citations: 6

- Single-center, interventional, open-label, proof-of-concept study
- 24 Pat. mit AT, mittleres Alter 17,5 Jahre
- NR 25mg/kg/Tag für 4 Mon. plus 2 Mon. wash-out

- Verbesserung in SARA und ICSAR nach 4 Monaten
- Verschlechterung nach Absetzen von NR

78 Short Communication

Nicotinamide Riboside for Ataxia Telangiectasia: A Report of an Early Treated Individual

Katja Steinbrücker¹ Elke Tiefenthaler¹ Eva-Maria Scherthaner¹ Julia Jungwirth¹
Saskia B. Wortmann^{1,2}

¹ University Children's Hospital, Paracelsus Medical University (PMU) Salzburg, Salzburg, Austria

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Neuropediatrics 2023;54:78–81.

GM2 Gangliosidose

RESEARCH ARTICLE

Efficacy and Safety of N-Acetyl-L-Leucine in Children and Adults With GM2 Gangliosidosis

Kyriakos Martakis, MD, Jens Claassen, MD, Jordi Gascon-Bayari, MD, Nicolina Goldschagg, MD, Andreas Hahn, MD, Anhar Hassan, MBCh, Anita Hennig, MD, Simon Jones, MD, Richard Kay, PhD, Heather Lau, MD, Susan Perlman, MD, Reena Sharma, MD, Susanne Schneider, MD, and Tatiana Bremova-Ertl, MD, PhD

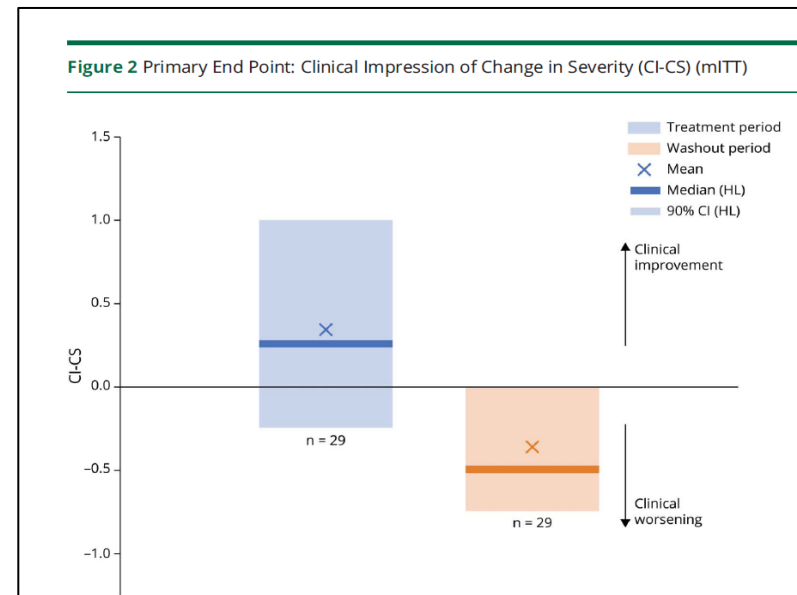
Neurology® 2023;100:e1072-e1083. doi:10.1212/WNL.000000000000201660

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- NALL sicher und effektiv
- Sign. Verbesserung der Funktion und QoL

- Phase IIb, open-label
- N=30 Patienten, Alter 6-55 Jahre
- Oral NALL für 6 Wo., danach 6 Wo. wash-out
- Prim. Zielparameter Clinical Impression of Change of Severity, Laufstrecke



GM2 Gangliosidose

RESEARCH ARTICLE

Efficacy and Safety of N-Acetyl-L-Leucine in Children and Adults With GM2 Gangliosidosis

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Neurology® 2023;100:e1072-e1083. doi:10.1212/WNL.000000000000201660

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