Revue de littérature
Thérapeutique

Pascal Cintas

CRMR neuromusculaire AOC

CHU Toulouse

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Hôpitaux de Toulouse

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Liens d’intérêt

- Consultant: LFB, Alnylam, Biogen, Roche

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Actualités 2021

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Actualités 2021

Gestion de la pénurie d'immunoglobulines

Gestion des fermetures de lits d'hospitalisation de jour

<u>Actualité thérapeutique quotidienne:</u>

- Réévaluer la dépendance thérapeutique
- Envisager les alternatives thérapeutiques

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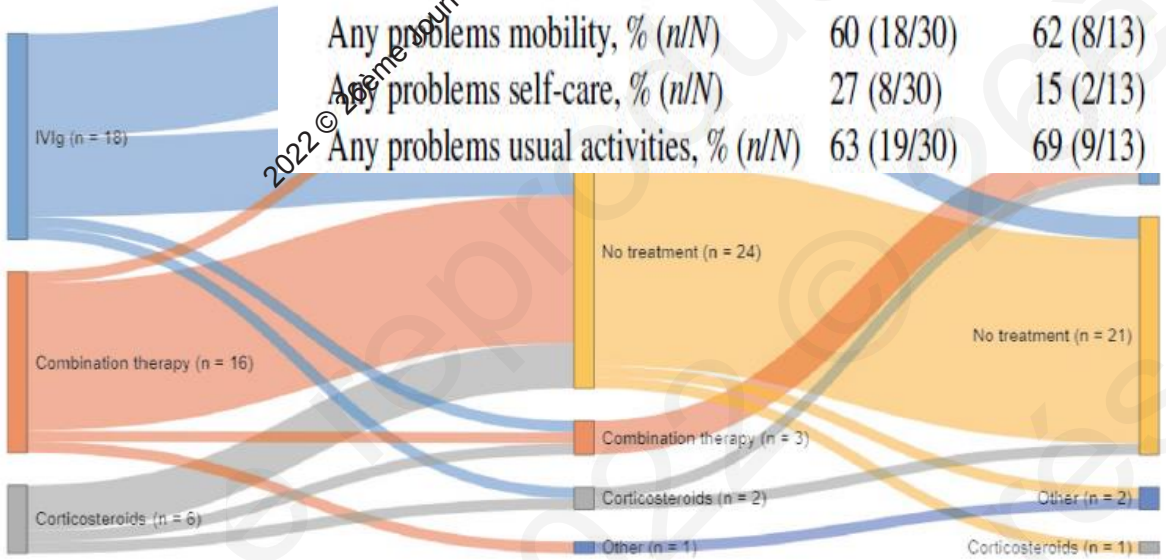
PIDC: dépendance thérapeutique dans la vraie vie

Clinical outcome of CIDP one year after start of treatment: a prospective cohort study

S. R. M. Bus[1] · M. C. Broers[2] · I. M. Lucke[1] · C. Bunschoten[2] · G. G. A. van Lieverloo[1] · M. E. Adriaense[1] · R. van Veen[1] · L. Wieske[1] · H. F. Lingsma[3] · H. S. Goedee[4] · W. L. van der Pol[4] · I. N. van Schaik[1,6] · P. A. Van Doorn[1] · B. C. Jacobs[2,5] · F. Eftimov[1] · for the ICOS Consortium

Erasmus MC	IgIV 2g/kg puis 0.4g/kg toutes les 2-4s
Amsterdam	IgIV 2g/kg puis 1g/kg + Methylprednisolone 1gr/3s pendant 18s
	6 mois (formes moins
	es 2-4s

	All treatment groups (n=40)	IVIg (n=18)	Corticosteroids[a] (n=6)	Combination treatment[b] (n=16)	Treated at 1 year (n=19)	Untreated at 1 year (n=21)
EQ-5D-5L						
Any problems mobility, % (n/N)	60 (18/30)	62 (8/13)	50 (2/4)	62 (8/13)	63 (10/16)	57 (8/14)
Any problems self-care, % (n/N)	27 (8/30)	15 (2/13)	25 (1/4)	39 (5/13)	13 (2/16)	43 (6/14)
Any problems usual activities, % (n/N)	63 (19/30)	69 (9/13)	25 (1/4)	69 (9/13)	56 (9/16)	71 (10/14)



			CS (n=6)
élément de suivi	89%	75%	50%
Rémission à 1 an	29%	44%	33%

- Taux important de pharmacodépendance IgIV
- Envisager traitements plus agressifs?

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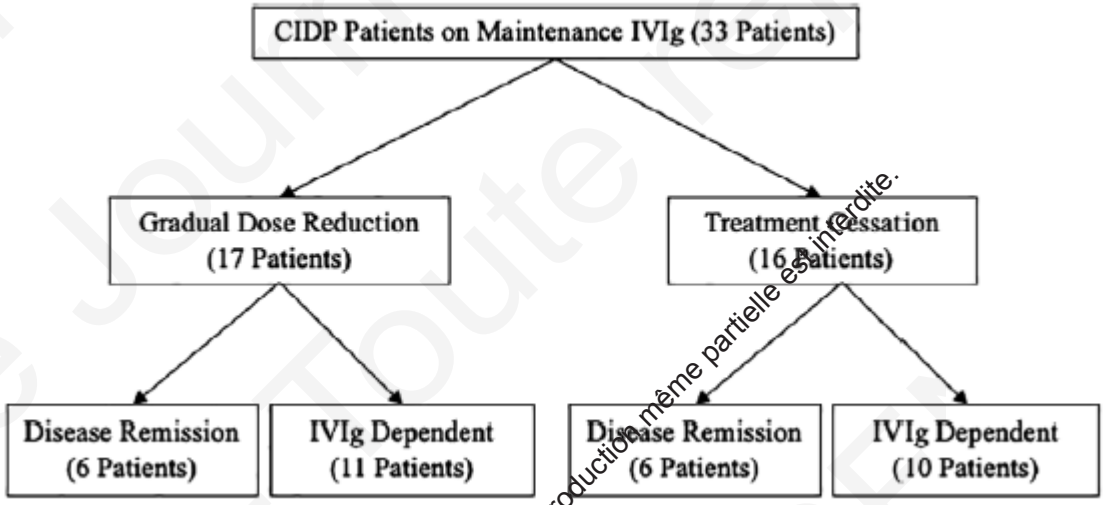
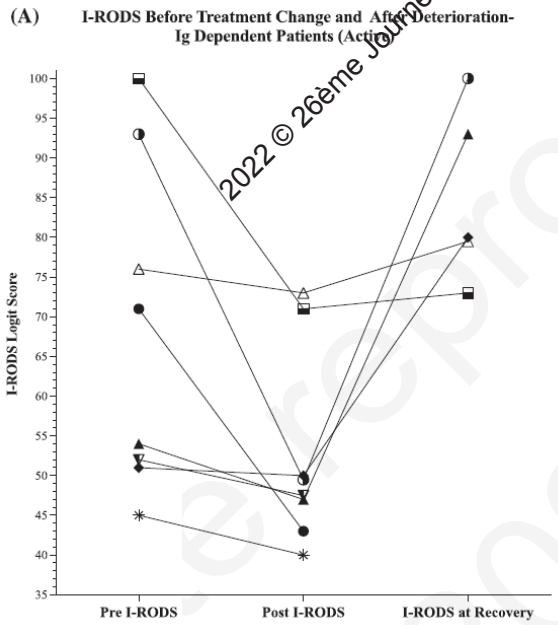
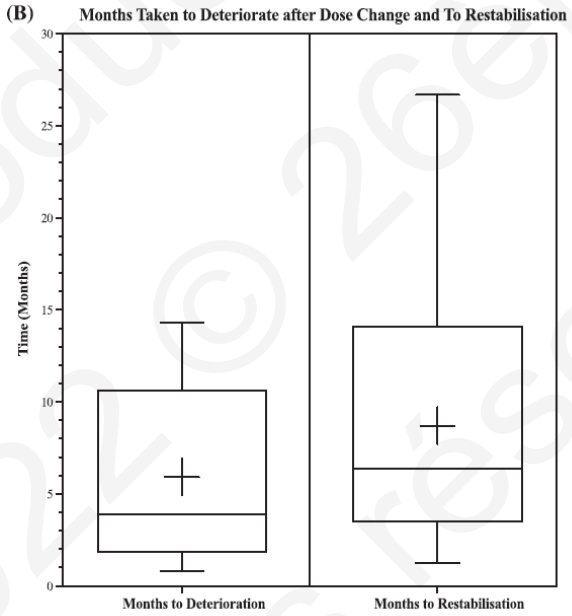
PIDC: dépendance thérapeutique dans la vraie vie

An approach to assessing immunoglobulin dependence in chronic inflammatory demyelinating inflammatory polyneuropathy

Mahima Kapoor[1,2] | Laura Compton[1] | Alex Rossor[1] | Elsbeth Hutton[2] | Hadi Manji[1] | Mike Lunn[1] | Mary Reilly[1] | Ailsling Carr[1]







Épreuve d'arret faisable:

- Magnitude de détérioration équivalente
- Restabilisation efficace
- Nécessité dans près de 50% des cas de doses un peu supérieures d'IgIV pour restabilisation

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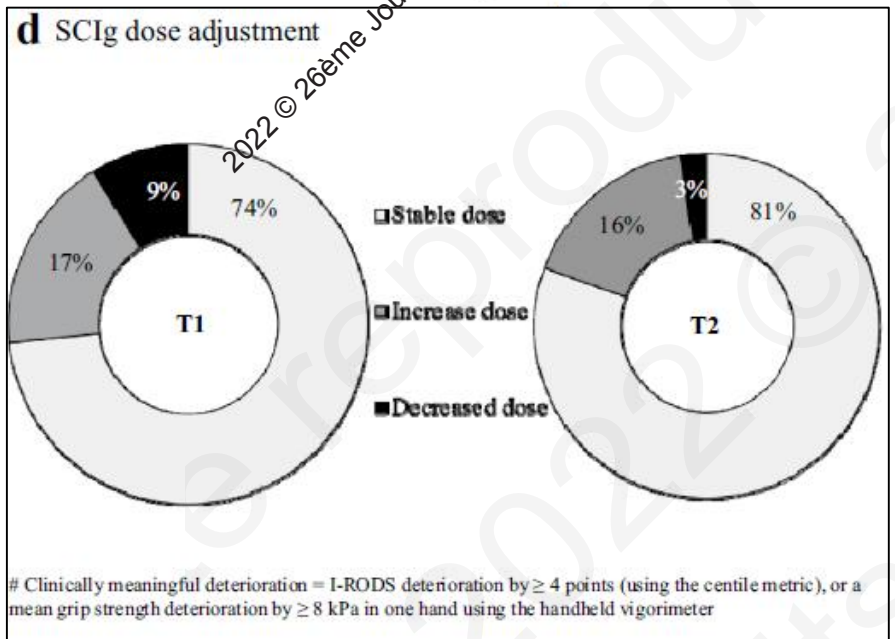
PIDC: dépendance thérapeutique dans la vraie vie

Subcutaneous immunoglobulin dose titration to clinical response in inflammatory neuropathy

Mahima Kapoor[1] · Ryan Keh[2] · Laura Compton[1] · Sarah Morrow[2] · David Gosal[2] · Hadi Manji[1] · Mary M. Reilly[1] · Michael P. Lunn[1] · Tim M. Lavin[2] · Aisling S. Carr[1]

	T = 0 IVIg maintenance		T = 1 12 months SCIg		T = 2 Last follow-up SCIg	
N	56		47		44	
Diagnosis	CIDP: 36	MMN:20	CIDP: 28	MMN:19	CIDP:25	MMN:19
I-RODS (logit scale)	61.3 (21.4) *34–94*	80.5 (20.4) *36–100*	59.7 (29.2) *20–96*	84.7 (17.5) *44–100*	66.0 (23.6) *22–96*	79.5 (22.8) *42–96*



Effet à long terme IgSC en relai IgIV:

- Maintenance de l'efficacité thérapeutique
- Nécessité d'augmentation des posologies fréquente chez 15-20% des patients/ dose IgIV

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PIDC: nodo-paranodopathies. Un modèle de résistance aux IgIV

Antibodies to the Caspr1/contactin-1 complex in chronic inflammatory demyelinating polyradiculoneuropathy

BRAIN 2021: 144; 1183–1196

Clinical and Laboratory Features in Anti-NF155 Autoimmune Nodopathy

Neurol Neuroimmunol Neuroinflamm 2022;9:e1098.

IgG$_1$ pan-neurofascin antibodies identify a severe yet treatable neuropathy with a high mortality

J Neurol Neurosurg Psychiatry 2021;**92**:1089–1095.

European Academy of Neurology/Peripheral Nerve Society guideline on diagnosis and treatment of chronic inflammatory demyelinating polyradiculoneuropathy: Report of a joint Task Force—Second revision

Disorders not classified as CIDP

Autoimmune nodopathies

Faible sensibilité aux IgIV

Sensibilité au Rituximab

IgG4-Mediated Neurologic Autoimmunities

Understanding the Pathogenicity of IgG4, Ineffectiveness of IVIg, and Long-Lasting Benefits of Anti–B Cell Therapies

Neurol Neuroimmunol Neuroinflamm 2022;9:e1116.

- Pas d'action de l'effet neutralisant en raison de leur isotype IgG1
- Pas d'action de l'inhibition du complément
- Peu d'action de l'effet d'inhibition des cytokines

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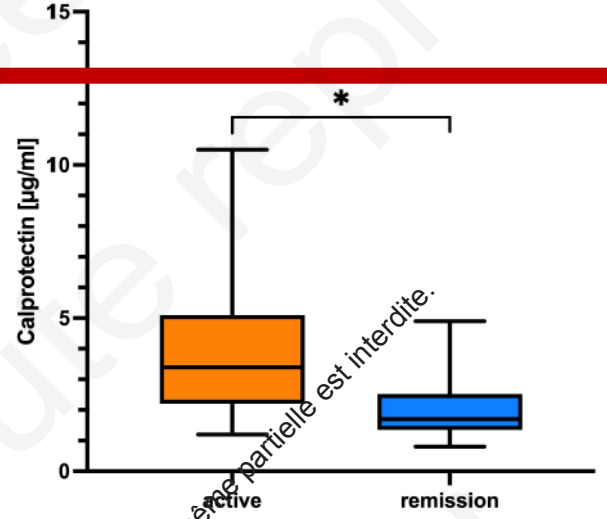
PIDC: PIDC: vers des biomarqueurs d'activité?

Prognostic value of nerve ultrasonography: A prospective multicenter study on the natural history of chronic inflammatory neuropathies

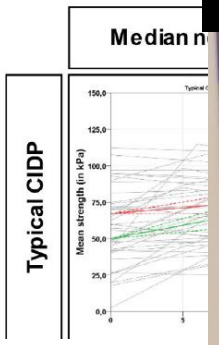
Eur J Neurol. 2021;00:1–12.

Calprotectin in Chronic Inflammatory Demyelinating Polyneuropathy and Variants—A Potential Novel Biomarker of Disease Activity

Frauke Stascheit[1,2†], Benjamin Hotter[1,2,3†], Sarah Klose[2], Christian Meisel[4,5], Andreas Meisel[1,2,3,6] and Juliane Klehmet[2,7]*



	Serum CLP	Serum NFL
	$r = 0.09$, $p = 0.47$	$r = 0.32$, $p = 0.006^{**}$
	$p = 0.025^{*}$	$p = 0.74$
	$r = 0.35$, $p = 0.013^{*}$	$r = -0.42$, $p = 0.0008^{***}$
	$r = -0.007$, $p = 0.95$	$r = 0.32$, $p = 0.009^{**}$



Absence d

Neuronopathies et AGO1 poster 17

Paramètres	AGO1	AGO1	P-valu
mRS chez SNN ayant reçu un traitement immunomodulateur			
mRS avant traitement - moyenne ± SEM	3.5 ± 0.3	2.4 ± 0.2	0.004
mRS après traitement - moyenne ± SEM	2.9 ± 0.3	2.5 ± 0.2	0.24
Réponse aux traitements - Diminution du mRS ≥1 - N (%)	7/13 (54%)	6/37 (16%)	0.02
Traitement de 1ère ligne seul	3/3 (100%)	4/18 (22%)	0.01
Traitement de 2ème ligne seul	1/1 (100%)	0/3 (0%)	0.08
Traitement de 1ère + 2ème lignes	3/9 (33%)	2/16 (13%)	0.22
IVIg	6/11 (55%)	4/22 (18%)	0.03
IVIg seul	1/1 (100%)	1/7 (14%)	0.08
IVIg avec autres traitements de 1ère ligne	2/2 (100%)	1/6 (17%)	0.049

- Pas d'effet prédictif de la réponse thérapeutique

- Calprotectine marqueur d'activité?
- NFL marqueur d'atteinte axonale?

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PIDC: quelle place du Rituximab?

Efficacy of rituximab treatment in chronic inflammatory demyelinating polyradiculoneuropathy: a systematic review and meta-analysis

Jianian Hu[1] · Chong Sun[1] · Jiahong Lu[1] · Chongbo Zhao[1] · Jie Lin[1]

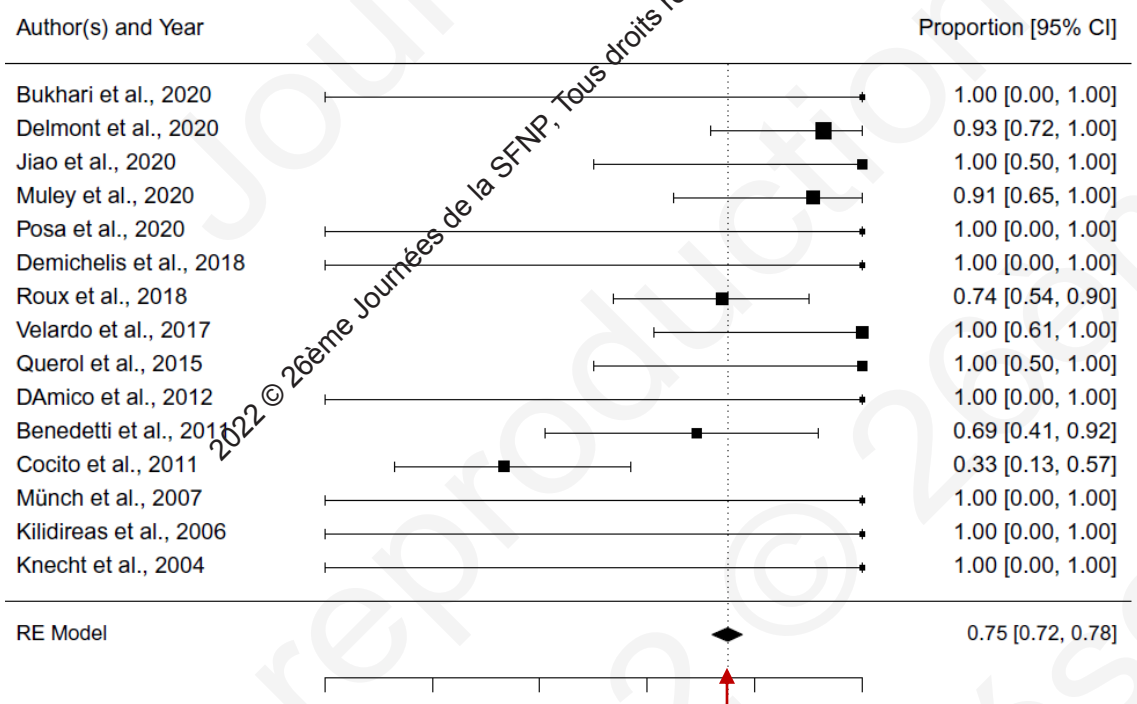

Fig. 2 Forest plot showing the pooled estimate of responsiveness of CIDP patients after rituximab therapy.

75% répondeurs

Seulement 72 patients analysables d'après ces études

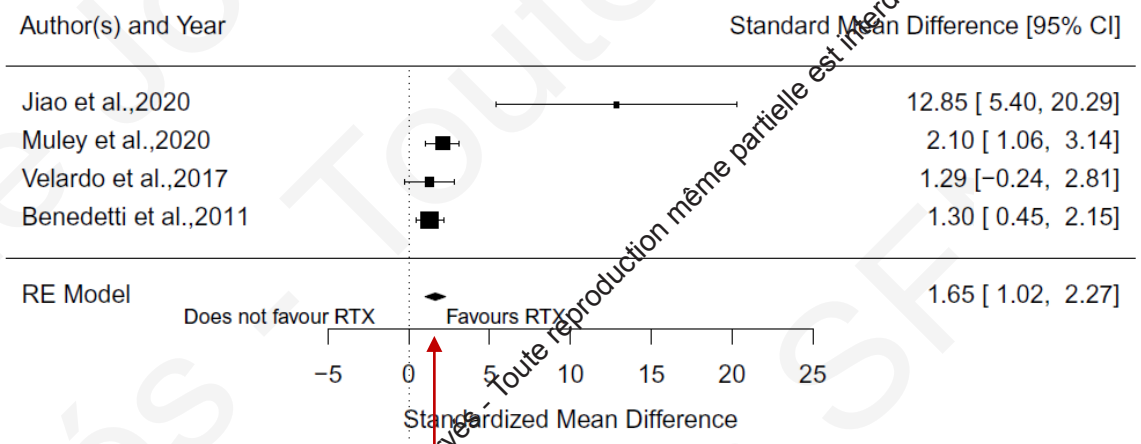

Fig. 3 Forest plot showing the improvement of INCAT score of CIDP patients after rituximab therapy

1.7pts INCAT

- Peu de données issues de la littérature
- Efficacité clinique convenue
- Place trop tardive dans la stratégie?

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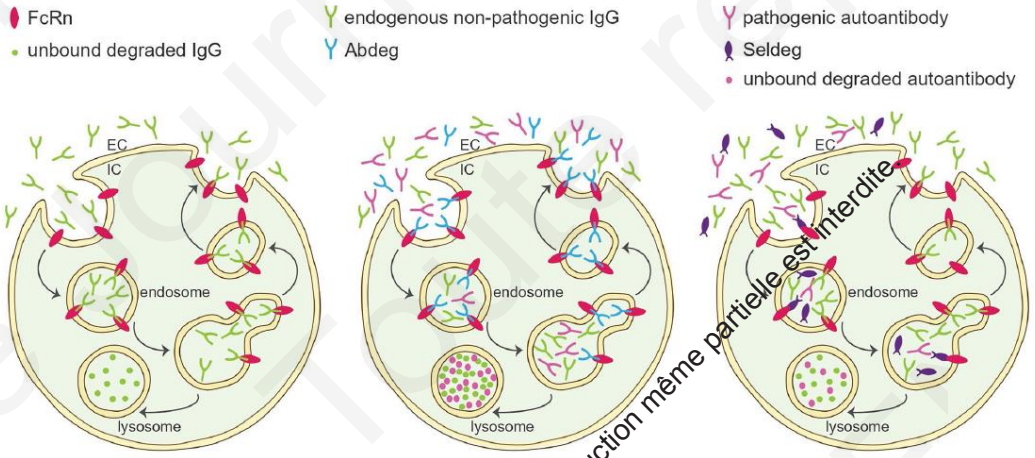
PIDC: les inhibiteurs des FcRN?

Neonatal Fc Receptor–Targeted Therapies in Neurology

Christopher Nelke[1] · Marianna Spatola[2] · Christina B. Schroeter[1] · Heinz Wiendl[3] · Jan D. Lünemann[3]



Treatment	Disease/Indication	Treatment strategy	Reference or ClinicalTrials.gov identifier
Efgartigimod (ARGX-113)	MG CIDP	Antagonism of FcRn (IgG1-derived Fc fragment with high affinity for FcRn)	[2, 78]
Nipocalimab	MG	Antagonism of FcRN (Abdeg, aglycosylated IgG1 mAb against FcRn)	NCT04951622 (recruiting)
Rozanolixizumab	MG CIDP	Antagonism of FcRn (Abdeg, humanized mAb against FcRn)	[80, 96]
Orilanolimab (SYNT001)	MG	Antagonism of FcRn (Abdeg, IgG4 mAb against FcRn)	[30]
CSL730/M230	Unknown	Antagonism of FcRn (Abdeg, IgG1 Fc multimer against FcRn)	NCT03375606 (terminated) NCT04446000 (ongoing)
Batoclimab (RVT-1401)	MG CIDP NMSOD	Antagonism of FcRn (Abdeg, mAb against FcRN)	[111] NCT04346888 (completed)

- Plusieurs études en cours
- Place de ces thérapies:
  - Phase initiale?
  - Au long cours?
  - Lors de rechutes?
- Impact sur les nodopathies IgG4?

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PIDC: place des autogreffes?

Hematopoietic stem cell transplantation for chronic inflammatory demyelinating polyradiculoneuropath

Richard K. Burt[1] · Roumen Balabanov[2] · Jinny Tavee[2] · X

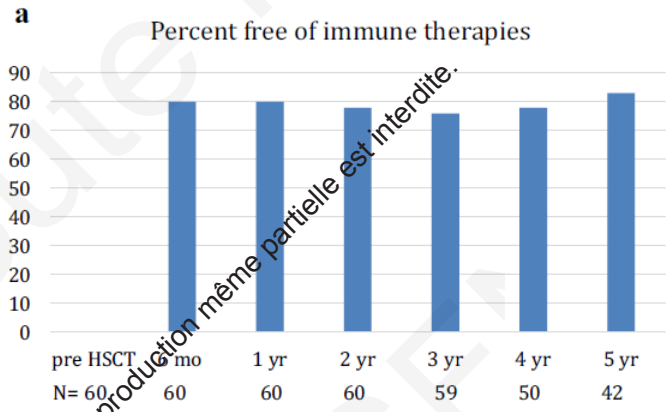
Prior treatment (number of patients)
Corticosteroids
Intravenous immunoglobulin (IVIG)
Plasmapheresis
Azathioprine
Mycophenolate mofetil
Rituximab
Cyclophosphamide
Methotrexate
Cyclosporine
Duration of symptoms before transplant: mean/medi

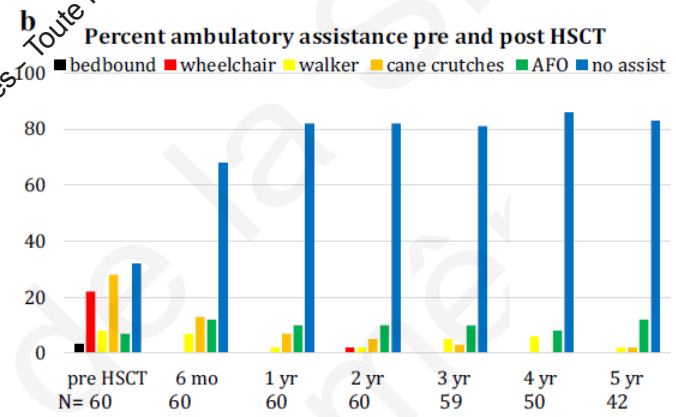
Inflammatory Neuropathy Cause and Treatment (INC
Rankin Score
Barthel Score
Rasch-built Overall Disability Scale (R-ODS) percentile
Overall Neuropathy Limitation Scale (ONLS)

Toxicities during and after transplantation	Number of patients
NCI grade 4 toxicity during transplantation	
Hypokalemia, BIBAP for dyspnea, TPN for nausea vomiting (one event each)	3/66 (4.5%)
NCI grade 3 toxicity during transplantation	
Hypophosphatemia	29/66 (44%)
Febrile neutropenia	26/66 (39%)
Hypokalemia	19/66 (29%)
Dyspnea (transient)	8/ 66 (12%)
Infections	8/66 (12%)
Clostridium difficile diarrhea	4 (6%)
Respiratory syncytial virus	1 (1.5%)
Atrial fibrillation	1/66 (3%)
Sinus tachycardia	1/66 (3%)
Deep venous thrombosis	1/66 (1.5%)
Syncope	1/66 (1.5%)
Allergic drug reaction	1/66 (1.5%)

Hematopoietic stem cell transplantation in chronic inflammatory demyelinating polyneuropathy: French experience about four patients, under the behalf of French society for bone marrow transplantation

Fanny Urbain[1] · Mathieu Puyade[2,3] · Céline Labeyrie[4,5] · Nicolas Maubeuge[6] · Angela Puma[7] · Pascal Cintas[8] · Gregory Pugnet[9,10] · Cristina Castilla-Llorente[11] · David Adams[4,5] · Dominique Farge-Bancel[1,12,13]





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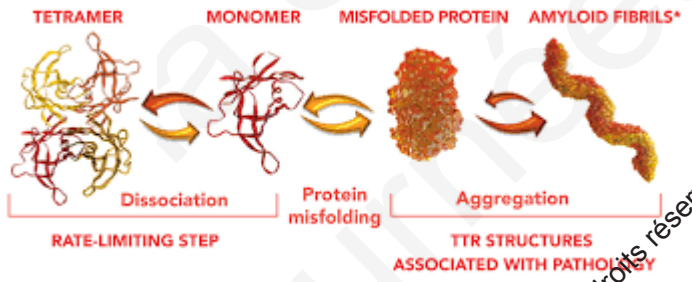
Amyloses ATTRv: quoi de neuf?



Tafamidis

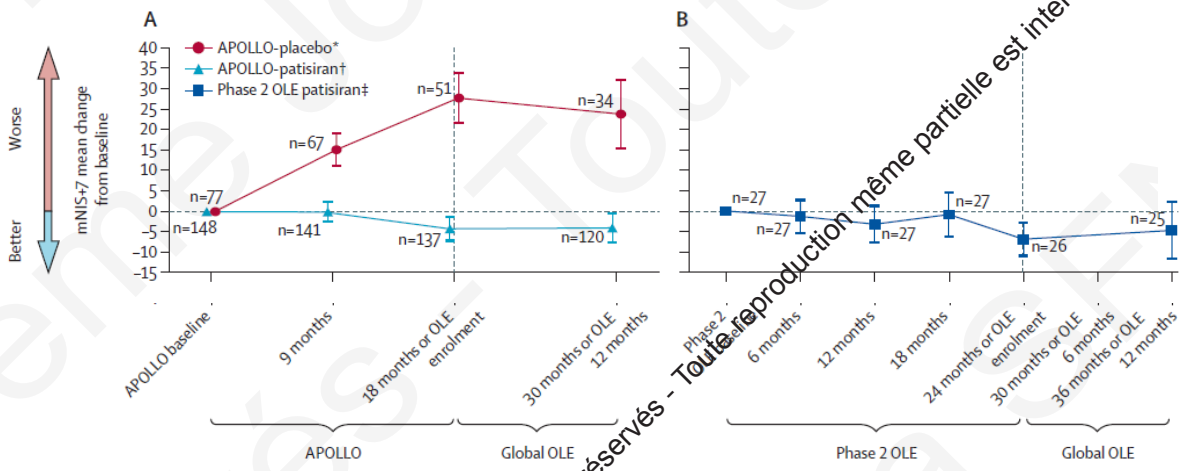
Patisiran

Inotersen

Maintien bénéfice à long terme?

Long-term safety and efficacy of patisiran for hereditary transthyretin-mediated amyloidosis with polyneuropathy: 12-month results of an open-label extension study

Lancet Neurol 2021; 20: 49–59

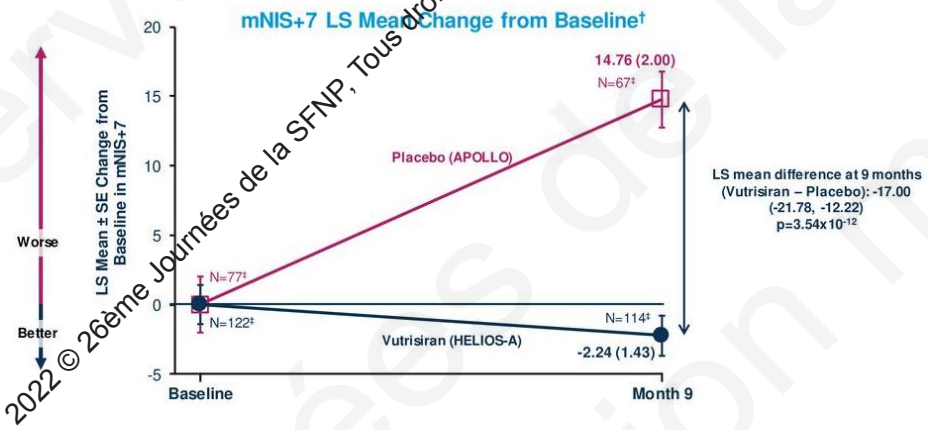


Améliorer efficacité et délivrance?

Eplotersen: SC/mois

En cours

Vutrisiran: SC/3mois



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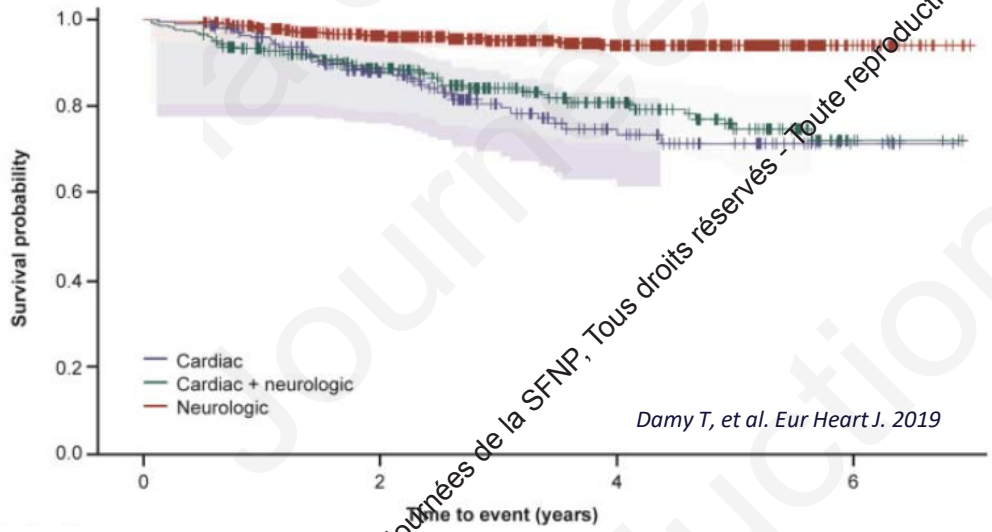
Amyloses ATTRv: quoi de neuf?



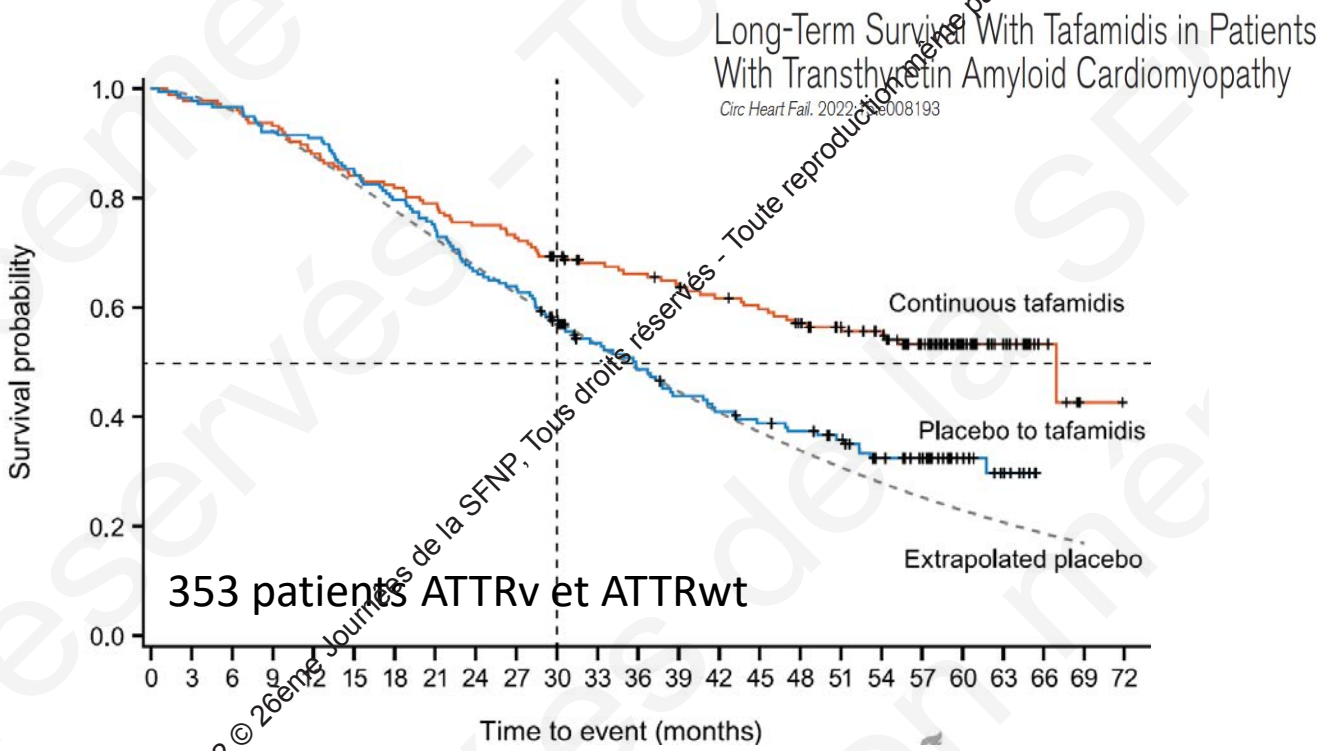
Damy T, et al. Eur Heart J. 2019

Enjeu sur la survie: l'atteinte cardiaque

- Nécessité du traitement précoce+++
- Impact Tafamidis sur atteinte cardiaque

Efficacité cardiaque des OAS et ARNi?:

- Apollo B, Helios B
- Cardio-TTRansform



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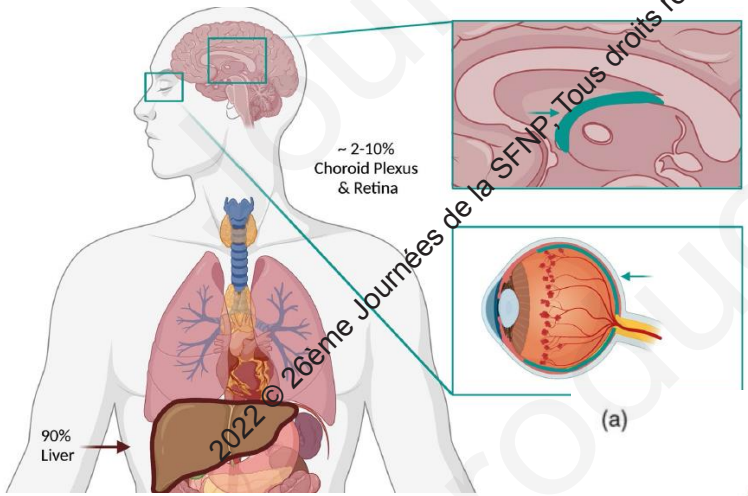
Amyloses ATTRv: quoi de neuf?

Are we creating a new phenotype? Physiological barriers and ethical considerations in the treatment of hereditary transthyretin-amyloidosis

Dohrn *et al. Neurol. Res. Pract.* (2021) 3:57



Transthyretin amyloid-related transitory events (TARTEs): Descriptive analysis of clinical, imagiological, and neurophysiological features

Journal of the Neurological Sciences 429 (2021) 118068

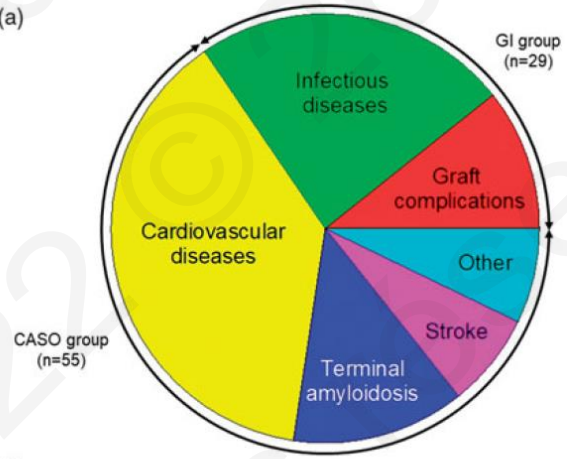
Age-dependent cognitive dysfunction in untreated hereditary transthyretin amyloidosis

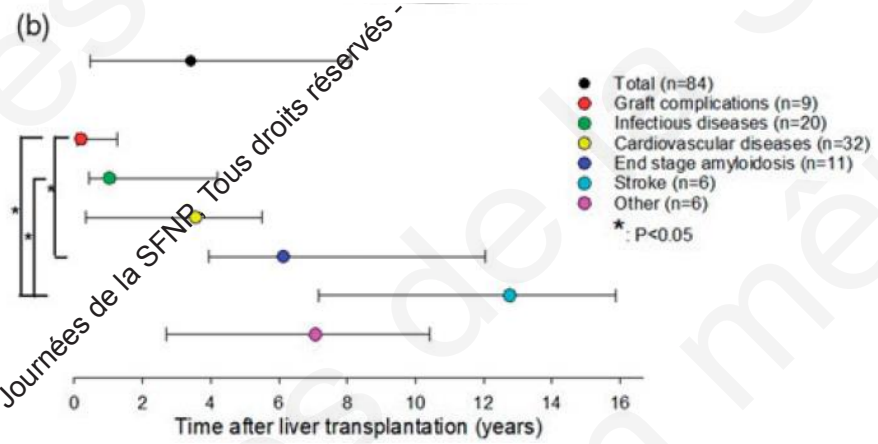
Journal of Neurology (2018) 265:299–307

Cerebral amyloid angiopathy in posttransplant patients with hereditary ATTR amyloidosis

Neurology® 2016;87:1-9

Cause of death analysis and temporal trends in survival after liver transplantation for transthyretin familial amyloid polyneuropathy





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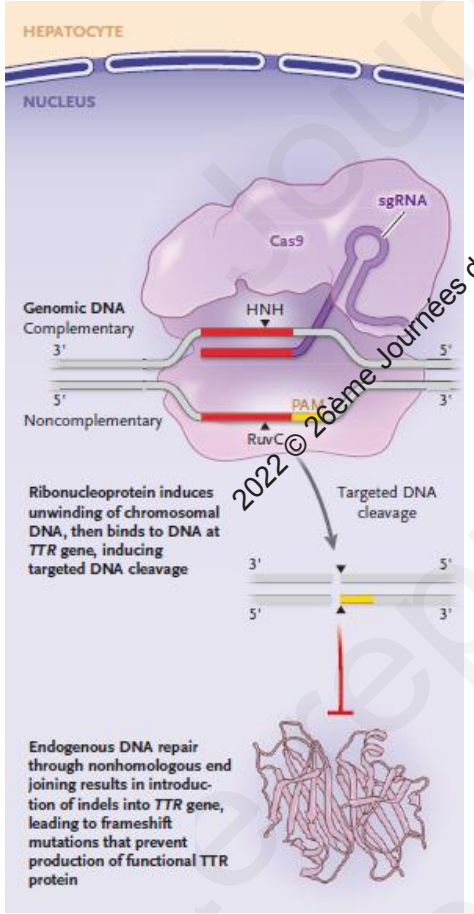
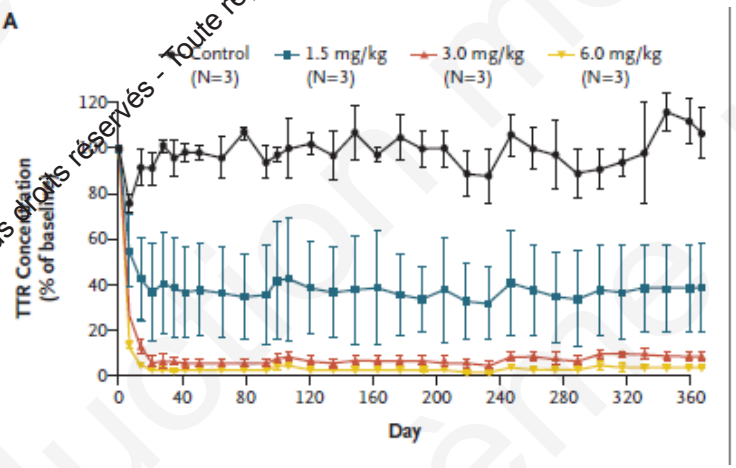
Amyloses ATTRv: quoi de neuf?

CRISPR-Cas9 In Vivo Gene Editing for Transthyretin Amyloidosis

N ENGL J MED 385;6

6 patients





- Preuve de principe de l'utilisation de l'édition du génome dans l'ATTR
- Nombreuses questions:
  - Effets off target?
  - Persistance effet au long cours
  - Impact clinique
- Ciblage purement hépatique: quel impact de la dépletion systémique en TTR sur production rétinienne et choroïdienne?

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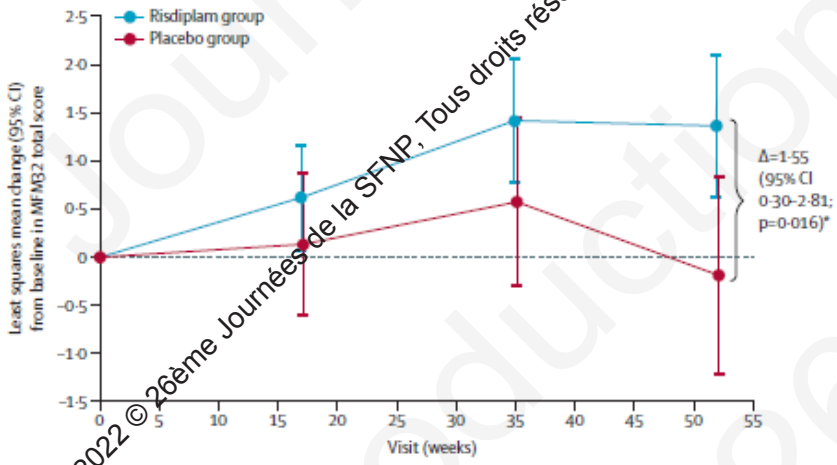
Amyotrophie spinale infantile: quoi de neuf chez l'adulte?

Safety and efficacy of once-daily risdiplam in type 2 and non-ambulant type 3 spinal muscular atrophy (SUNFISH part 2): a phase 3, double-blind, randomised, placebo-controlled trial

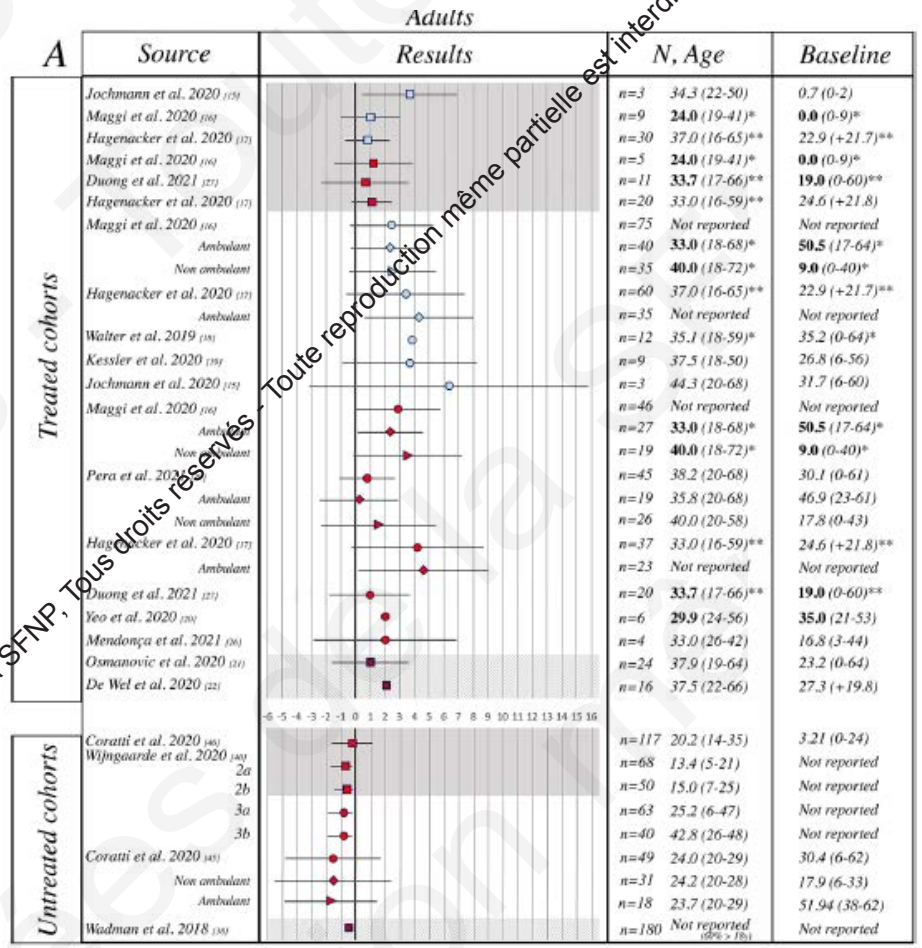
www.thelancet.com/neurology Vol 21 January 2022



Subgroup	Category	(N* Risdiplam, N* Placebo)	Estimate	[95% CI]	P-value
Overall		(120, 60)	0.58	[-0.53, 1.69]	0.3015
Age groups	2 – <6	(37, 18)	0.23	[-1.78, 2.24]	0.8189
	6 – 11	(39, 18)	1.25	[-0.71, 3.20]	0.2099
	12 – 17	(30, 16)	0.74	[-1.42, 2.91]	0.4988
	18 – 25	(14, 8)	-0.73	[-3.78, 2.32]	0.6364

Motor function in type 2 and 3 SMA patients treated with Nusinersen: a critical review and meta-analysis

Coratti *et al. Orphanet J Rare Dis* (2021) 16:430



- Attente des premières données de vie réelle adulte du Risdiplam
- Intérêt des RCP nationales

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Amyotrophie spinale infantile: quoi de neuf chez l'adulte?

Nusinersen treatment in adults with severe spinal muscular atrophy: A real-life retrospective observational cohort study

C. Lefeuvre [a,b,g,*,1,5], M. Brisset [a,b,g,1,5], M. Sarlon [c,g,2], N. Petit [a,g,2], D. Orlikowski [e,i,g,2], B. Clair [e,g,2], T. Thiry [f,g,2], R.-Y. Carlier [d,f,g,2], H. Prigent [b,g,h,2], G. Nicolas [a,b,d,g,2], D. Annane [e,g,2], P. Laforet [a,b,d,g,3], S. Pouplin [c,d,g,4]

Number of patients	18
Females/Males	6/12
Baseline age, years, mean ± SD (range)	28.0 ± 5.9 (21–39)
SMA type, *n* (%)	
1	2 (11.1)
1bis	5 (27.8)
2	9 (50.0)
3	2 (11.1)

	MFM-32		
	D2	D3	Total
All participants, *n* = 18			
Baseline			
Day 0	3.5 [2.0; 7.5]	8.0 [3,25; 11,75]	12.0 [4.0; 18.0]
Difference in total score			
Days 0–63	0.0 [0.0; 1.0]	0.25 [−0.75; 1.0]	0.25 [0.0; 2.75]
Days 0–183	0.5 [−1.0; 1.75]	1.0 [0.0; 2.75]	0.5 [−0.75; 4.25]
Days 0–303	0.0 [−0.75; 1.0]	0.0 [−1.0; 1.0]	0.0 [−0.75; 1.75]

Table 3 – Median [interquartile range] Canadian Occupational Performance Measure scores for performance and satisfaction in the overall population and in the subgroups defined by Motor Function Measure (MFM)-32 scores < or ≥ the mean (15.6%) in the total population.

	COPM	
	Performance	Satisfaction
All participants, *n* = 18		
Baseline		
Day 0	19.0 [13.75; 25.0]	20.5 [13.5; 24.5]
Difference in total score		
Days 0–63	0.5 [0.0; 3.75]	0.0 [−1.75; 5.75]
Days 0–183	1.5 [0.0; 3.75]	2.0 [2.0; 8.0]**
Days 0–303	0.5 [0.5; 3.0]	2.0 [2.0; 4.0]
Low-MFM group, *n* = 10		
Baseline		
Day 0	18.0 [13.25; 22.75]	20.5 [13.5; 21.0]
Difference in total score		
Days 0–63	1.0 [0.0; 3.75]	0.0 [−1.5; 4.5]
Days 0–183	1.5 [0.0; 3.75]	1.5 [−0.75; 8.0]
Days 0–303	0.5 [0.0; 3.0]	1.5 [0.0; 4.0]
High-MFM group, *n* = 8		
Baseline		
Day 0	25.5 [15.25; 27.5]	21.0 [14.5; 26.0]
Difference in total score		
Days 0–63	0.5 [−2.0; 3.5]	2.5 [−1.25; 5.5]
Days 0–183	1.5 [0.0; 4.0]	2.5 [0.75; 5.75]
Days 0–303	1.0 [−0.25; 5.0]*	2.5 [−0.25; 5.5]

* P = 0.01.
** P = 0.02.

- Formes sévères à très sévères adultes
- Effets significatif sur MCRO sur rendement et satisfaction
- Nécessité d'évaluer les biomarqueurs particulièrement pour formes tardives:
  - ➢ études SPIN2, SMOB

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CMT: CMT1A

A double-blind, placebo-controlled, randomized trial of PXT3003 for the treatment of Charcot–Marie–Tooth type 1A

Shahram Attarian[1*], Peter Young[2], Thomas H. Brannagan[3], David Adams[4], Philip Van Damme[5,6],

Baclofene
Sorbitol
Naltrexone

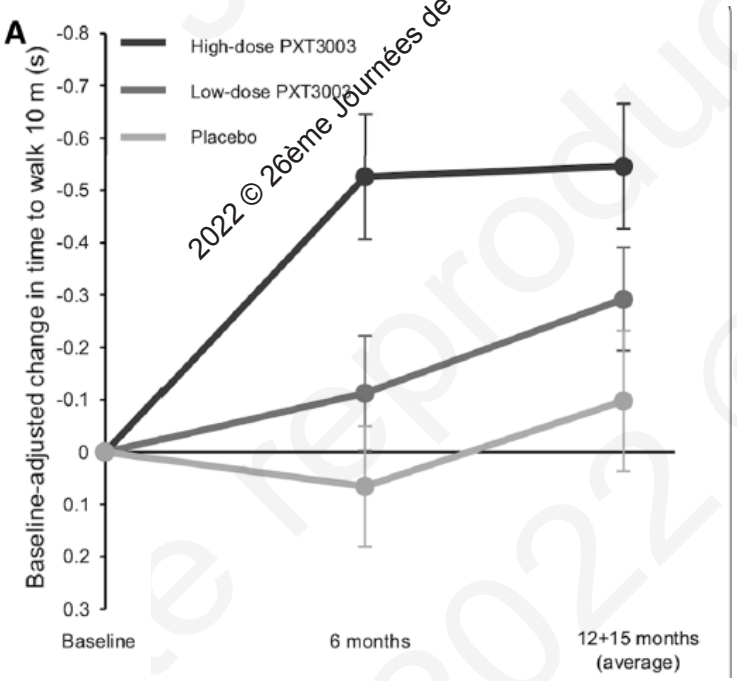

Table 2 Treatment effect of high and low-dose PXT3003 vs placebo in exploratory efficacy endpoints (mFAS)

	Test	High-dose PXT3003	Low-dose PXT3003
ONLS	Arm score (↓)	−0.19 (−0.43, 0.04)	0.01 (−0.19, 0.22)
	Leg score (↓)	**−0.18** (−0.36, 0.01)	−0.13 (−0.29, 0.02)
QMT	Grip strength (↑) (kg)	−1.49 (−3.92, 0.93)	0.37 (−1.64, 2.37)
	Dorsiflexion strength (↑) (Newton)	7.48 (−3.60, 18.[illegible])	4.09 (−5.01, 13.20)
Electrophysiology	CMAP (↑) (mV)	−0.33 (−1.05, [illegible].40)	0.14 (−0.46, 0.74)
	Motor nerve conduction velocity (↑) (m/s)	0.08 (−1.[illegible]2, 1.67)	−0.09 (−1.42, 1.25)
	Distal motor latency (↓) (ms)	−0.20 (−1.58, 1.18)	0.08 (−1.07, 1.24)
	Radial SNAP (↑) (μV)	−0.[illegible]1 (−1.20, 0.99)	−0.04 (−0.96, 0.88)
Quality of life	Mobility (↓)	−0.07 (−0.35, 0.20)	−0.11 (−0.34, 0.12)
	Self-care (↓)	−0.12 (−0.31, 0.06)	**−0.18** (−0.34, −0.03)
	Usual activities (↓)	−0.02 (−0.27, 0.23)	−0.11 (−0.32, 0.10)
	Pain/discomfort (↓)	0.00 (−0.25, 0.26)	−0.12 (−0.33, 0.09)
	Anxiety/depression (↓)	−0.04 (−0.29, 0.22)	−0.06 (−0.28, 0.15)
	Visual analog scale (↑) (mm)	2.57 (−2.36, 7.50)	**4.67** (0.59, 8.75)

Essai encourageant mais problème de cristallisation dans bras à forte dose.
→ Nouvelle phase 3 en cours

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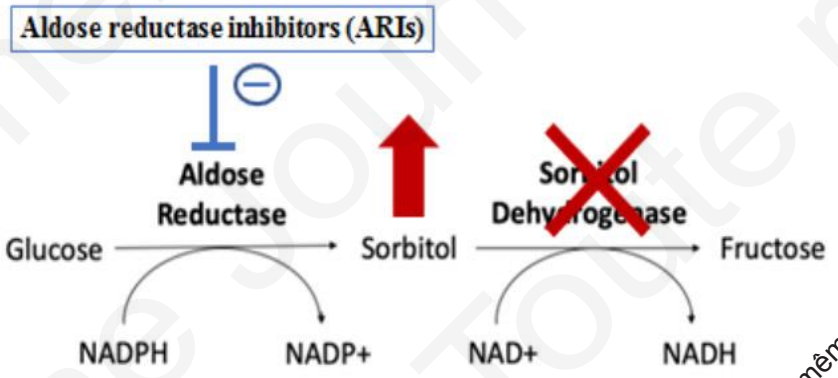
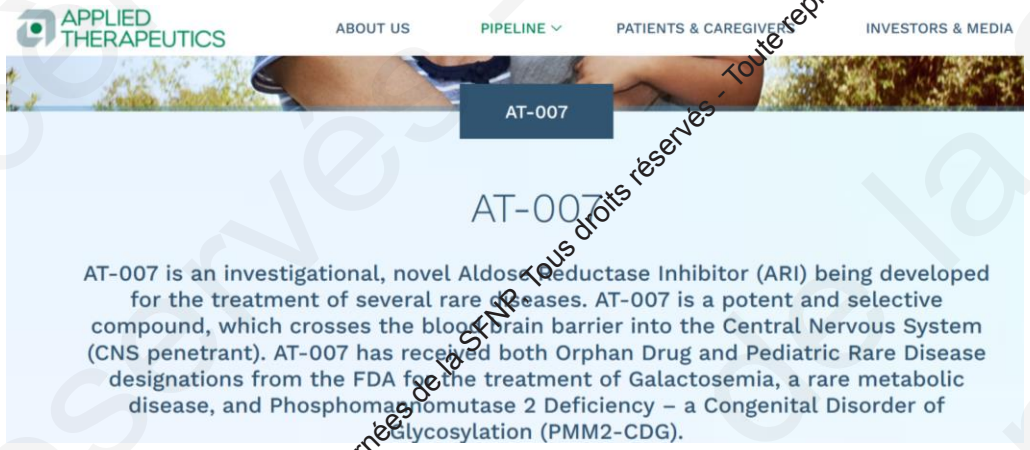
CMT: un nouveau CMT2

BIALLELIC MUTATIONS IN *SORD* CAUSE A COMMON AND POTENTIALLY TREATABLE HEREDITARY NEUROPATHY WITH IMPLICATIONS FOR DIABETES

Nat Genet. 2020 May ; 52(5): 473–481.

	N=45
Male	32 (71%)
Age of onset (years)	17 ± 8 (2–40)
Age at examination (years)	34 ± 13 (15–70)
Family history of neuropathy	14 (31%)
CMT subtype	
CMT2	23 (51%)
dHMN	18 (40%)
CMT intermediate	4 (9%)
Foot deformities	31 (69%)
Upper limb weakness	
Proximal muscle groups	0/44 (0%)
Distal muscle groups	26/44 (59%)
Lower limb weakness	
Proximal muscle groups	2/44 (5%)
Distal muscle groups	43/44 (98%)
Prominent involvement of foot plantar flexion*	15/37 (41%)
Reduced vibratory sensation	17/40 (42%)
Reduced pinprick superficial sensation	13/39 (33%)
Disease severity	
Mild	30 (67%)
Moderate	14 (31%)
Severe	1 (2%)
Use of ankle-foot orthoses (AFOs)	19 (42%)
Other walking aids	2 (4%)
Nerve conduction study	
Reduced motor conduction velocity	11/42 (26%)
Reduced sensory action potentials	26/40 (65%)





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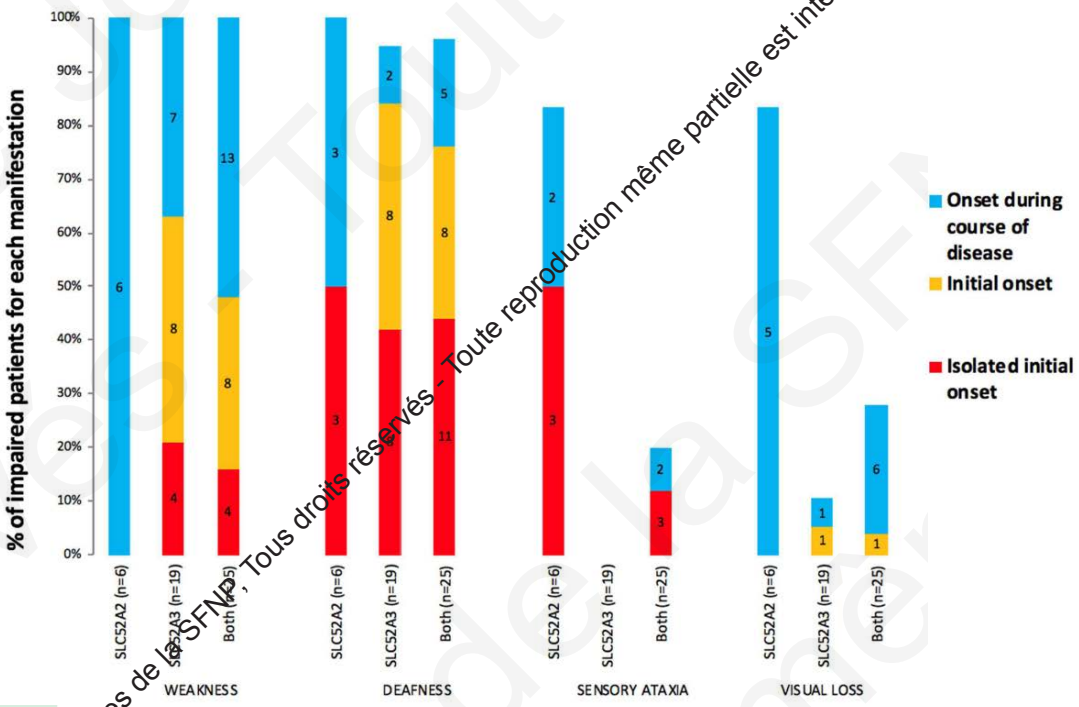
Une autre neuropathie traitable: Brown Vialetto Van Laere

Late-onset riboflavin transporter deficiency: a treatable mimic of various motor neuropathy aetiologies

Christophe Carreau,[1] Charline Benoit,[2] Guido Ahle,[3] Cécile Cauquil,[4] Agathe Roubertie,[5] Timothée Lenglet,[6] Jeremy Cosgrove,[7] Isabelle Meunier,[8] Alice Veauville-Merllié,[9] Cécile Acquaviva-Bourdain,[9] Yann Nadjar[1]

Table 1 Demographic, clinical, biological and electrophysiological data of the pooled cohort (25 patients)

	SLCA52A2	*SLCA52A3*	Total
No of patients	6 (24%)	19 (76%)	25 (100%)
Sex ratio (M/F)	3/6 (50%)	3/19 (21%)	6/25 (24%)
Consanguinity	4/5 (80%)	4/14 (29%)	8/19 (42%)
Age at onset (years) (n=25)	5.25±2.63 (3–8)	17±7.59 (8–33.5)	15±8.38 (3–33.5)
Age at MN onset (years) (n=25)	13±4.49 (10–20)	18±8.53 (10–35)	16.8±7.99 (10–35)
Age at last follow-up (years) (n=24)	22±5.27 (17–32) (n=6)	30±11.32 (15–57) (n=18)	28±10.73 (15–57)
Years from first symptom to last follow-up (n=12)	18±3.76 (14–24) (n=6)	14±11 (0.5–37) (n=18)	15±9.46 (0.5–37)
Death	1 (17%)	1 (5%)	2 (8%)
Response to riboflavin treatment			
No	0/4 (0%)	2/10 (20%)	2/14 (14%)
Mild	2/4 (50%)	3/10 (30%)	5/14 (36%)
Strong	2/4 (50%)	5/10 (50%)	7/14 (50%)
Follow-up after treatment onset	1.25±1.5 (0–3)	1.6±1.9 (0–6)	1.5±1.73 (0–6)
Electrophysiological tests			
Normal EMG/NCS	0/6 (0%)	0/17 (0%)	0/23 (0%)
Pure motor defects EMG/NCS	1/6 (17%)	17/17 (100%)	17/23 (74%)
Pure sensory defects EMG/NCS	0/6 (0%)	0/17 (0%)	0/23 (0%)
Motor and sensory defects EMG/NCS	5/6 (83%)	0/17 (0%)	5/23 (22%)
Impaired auditory-evoked potentials	1/1 (100%)	11/11 (100%)	12/12 (100%)
Impaired visual-evoked potentials	1/1 (100%)	1/6 (17%)	2/7 (29%)
Acylcarnitine profile			
Normal	1/4 (25%)	5/8 (62%)	6/12 (50%)
MADD	3/4 (75%)	2/8 (25%)	5/12 (42%)
Abnormal not MADD	0/4 (0%)	1/8 (12%)	1/12 (8%)
Blood level riboflavin			
Normal	1/1 (100%)	3/3 (100%)	4/4 (100%)
Abnormal	0/1 (0%)	0/3 (0%)	0/4 (0%)



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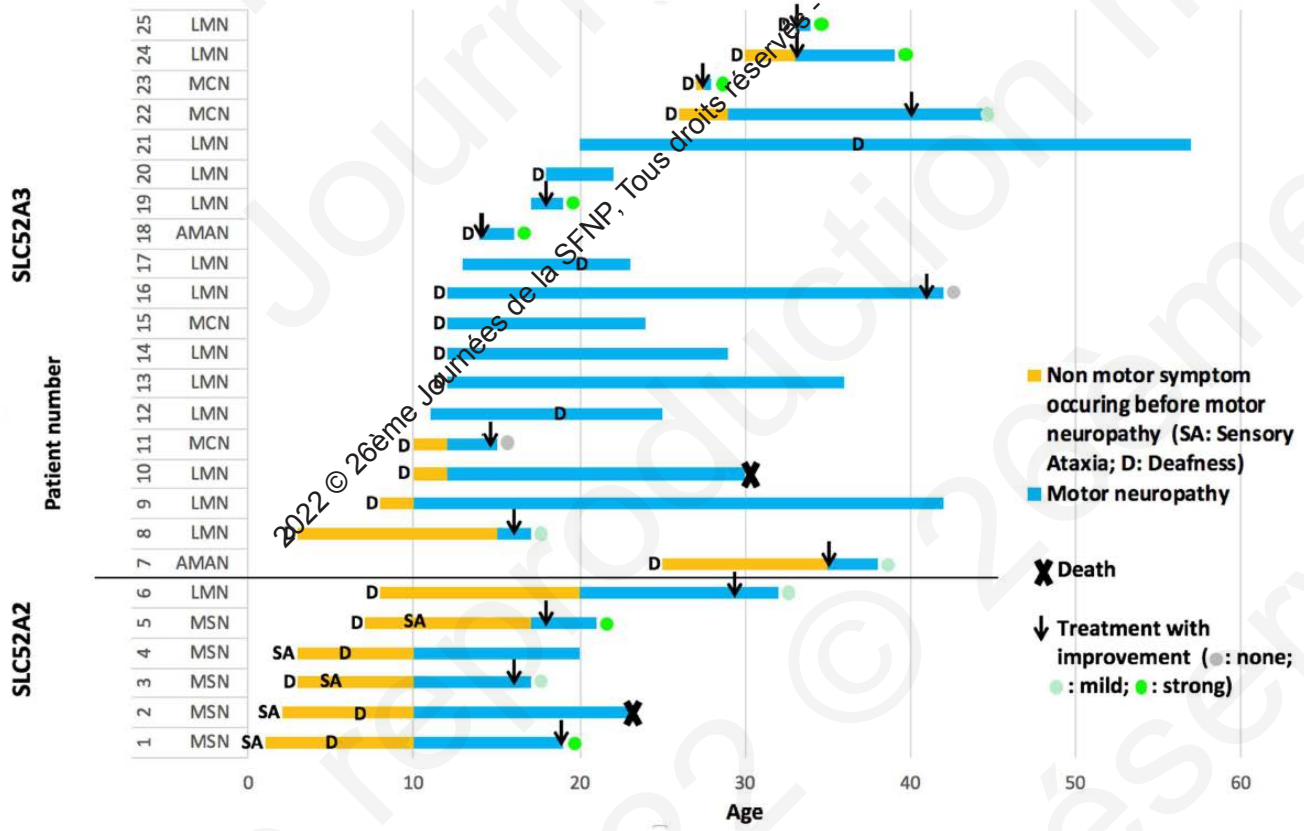
Une autre neuropathie traitable: Brown Vialetto Van Laere



- Présence de surdité quasi systématique à l'entrée dans la maladie motrice
- Début brutal (4%) ou subaigu (24%):
  - Guillain barré like
  - Pseudomultinévrite
  - Motoneurone périphérique

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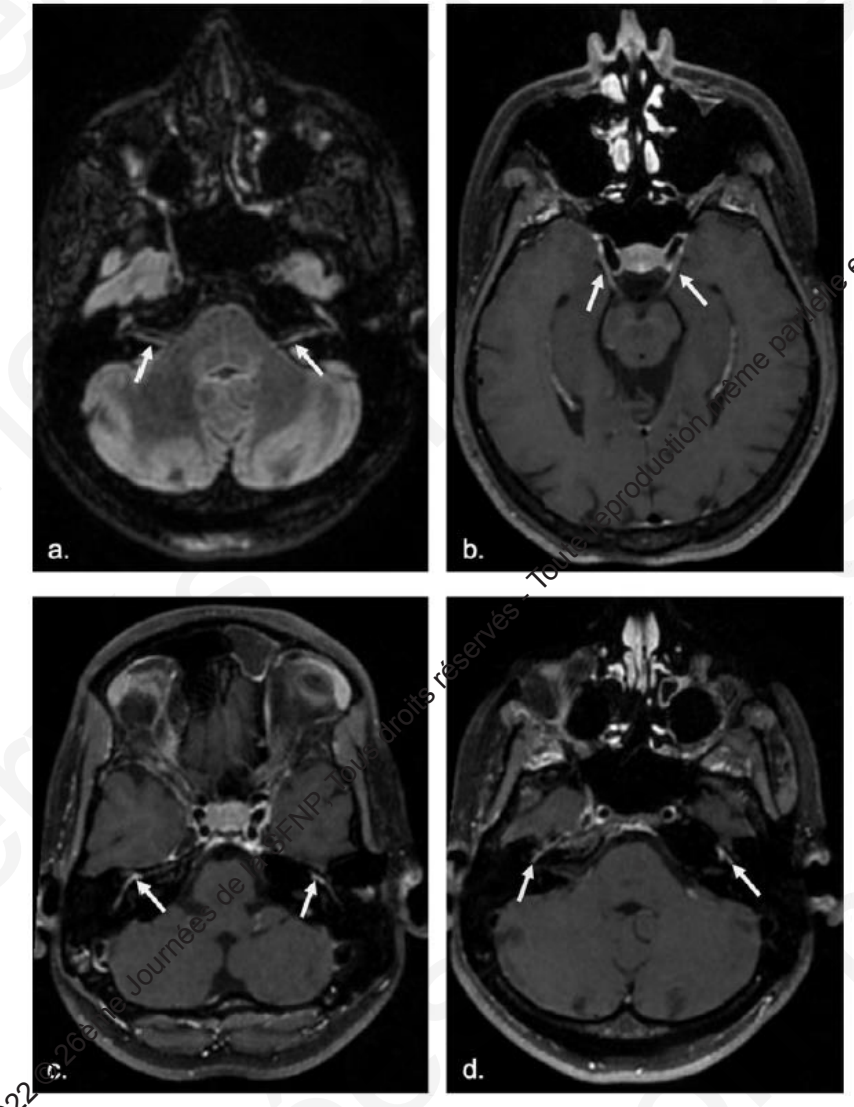
Une autre neuropathie traitable: Brown Vialetto Van Laere

Three cases of adult-onset Brown-Vialetto-Van Laere syndrome: Novel variants in *SLC52A3* gene and MRI abnormalities

Guillaume Carey[a,c,*], Gregory Kuchcinski[a,d], Fanny Gauvrit[f], Luc Defebvre[a,c], Sylvie Nguyen[b,e], Claire-Marie Dhaenens[a], Anne Frédérique Dessein[a], Chrisjme Vianey-Saban[g], Cécile Acquaviva[g], Céline Tard[a,b,c]



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Conclusion: propositions pour la présentation SFNP 2023

- Bouleverse dies neuromusc
  - Nouvelles nir précisement (inhibiteu
  - Nouvelles ns certaines populatio
  - Nouvelles RNi et stabilisate
  - Ancienne ques agressives, nouveaux biomarqueurs, gestion des igrv…

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