MaRDi
Maladies Rares Digestives

Avancées dans la compréhension des POIC

Intégration des données génétiques et histologiques

2007-2023

Minh-Chau Ta

1958

Dudley et al.

Intestinal pseudo-obstruction

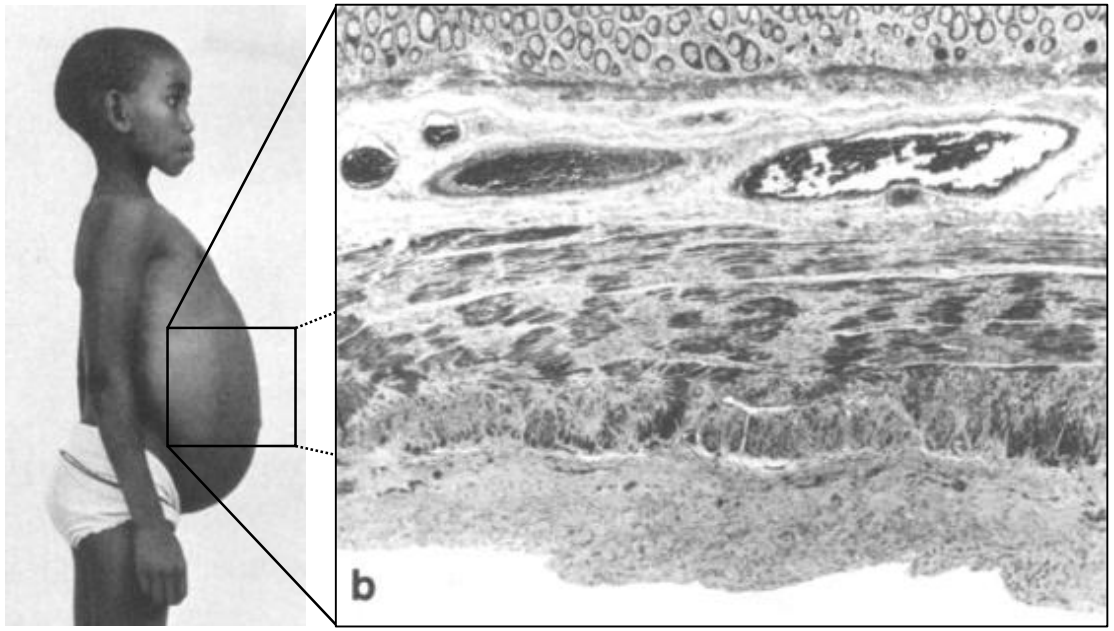
THE ROYAL COLLEGE OF SURGEONS OF EDINBURGH

1992

Rode et al.

Degenerative Leiomyopathy

Pediatric Surgery International



2010

Knowles et al.

The London classification

- **Myopathy**
- **Neuropathy**
- **Mesenchymopathy**

Gut

2012

Lehtonen et al.

ACTG2

Gastroenterology

2022-2024

CLINIQUE

+350 variables

doqboard

GENETIQUE

Sang

Tissus congelés

Sanger
Panel NGS
Exome
Genome

Necker ENFANTS MALADES HÔPITAL UNIVERSITAIRE

SEQOIA Médecine génomique

institut imagine GUÉRIR LES MALADIES GÉNÉTIQUES

HISTOLOGIE

Biopsies chirurgicales résections

Intestin grêle, côlon

HES, PAS
Picrosirius
IHC:

- Actine
- Desmine
- Hu C/D
- CD117

Hôpital Beaujon AP-HP

Âge médian au début des symptômes

6 ans

Âge médian au diagnostic

22 ans

1ère hospitalisation	Tous n=130	Gastro-pédiatrie n=71	Gastroentérologie adulte n=59
Obstruction fonctionnelle	82 (63 %)	57 (80 %)	25 (42 %)
Volvulus	12	2	10
Diarrhée sévère	15	3	12
Constipation sévère	14	6	8
Vomissements	7	3	4

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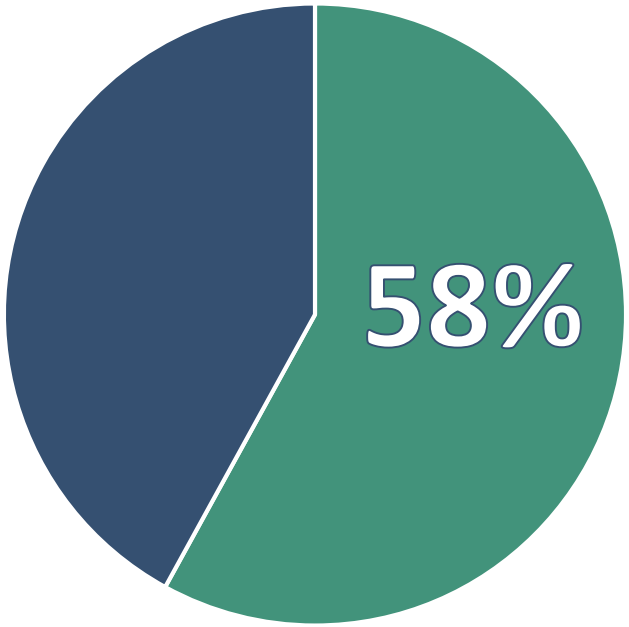
- **Inertie colique** (n=24)
- **Anorexie mentale** (n=20)

- **Gastroentérite aiguë** (n=5)
- **Maladie de Crohn** (n=4)
- **Lymphome/Myélome** (n=3)

Tests génétiques, n=112



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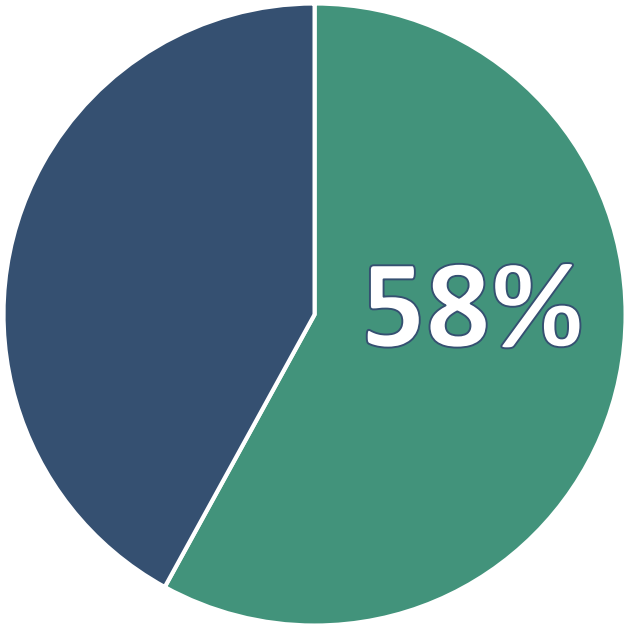


Targeted NGS long-range PCR

ACTG2 n=29
MYH11 n=9
SGO1 n=2
FLNA n=2

STAT1 n=1
SOCS1 n=1

TYMP n=9
POLG n=3
MT-TL1 n=3
MT-ATP6 n=1
MT-TV n=1
mt-DNA^{DEL} n=2

Exome sequencing

TTC37 n=1

Genome sequencing

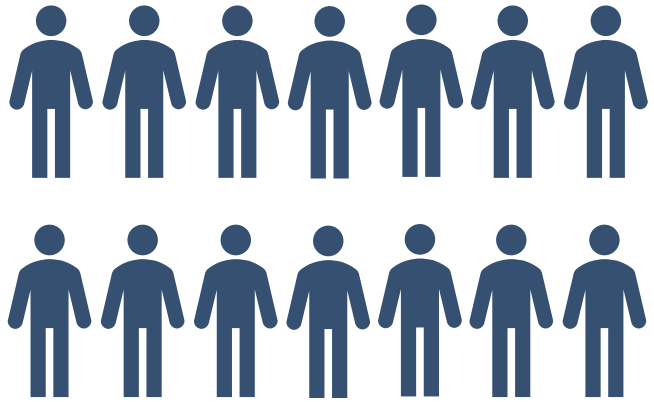
POGZ n=1

Richards S et al. Genet Med. 2015 May;17(5):405-24.

Tests génétiques, n=112

1ère obstruction après

50 ans (n=14)



ACTG2 n=29
MYH11 n=9
SGO1 n=2
FLNA n=2

STAT1 n=1
SOCS1 n=1

TTC37 n=1

TYMP n=9
POLG n=3
MT-TL1 n=3
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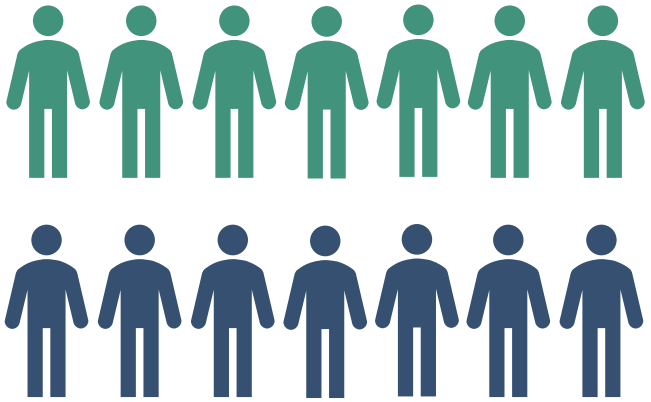

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Tests génétiques, n=112

« Anorexia nervosa »
(n=20)

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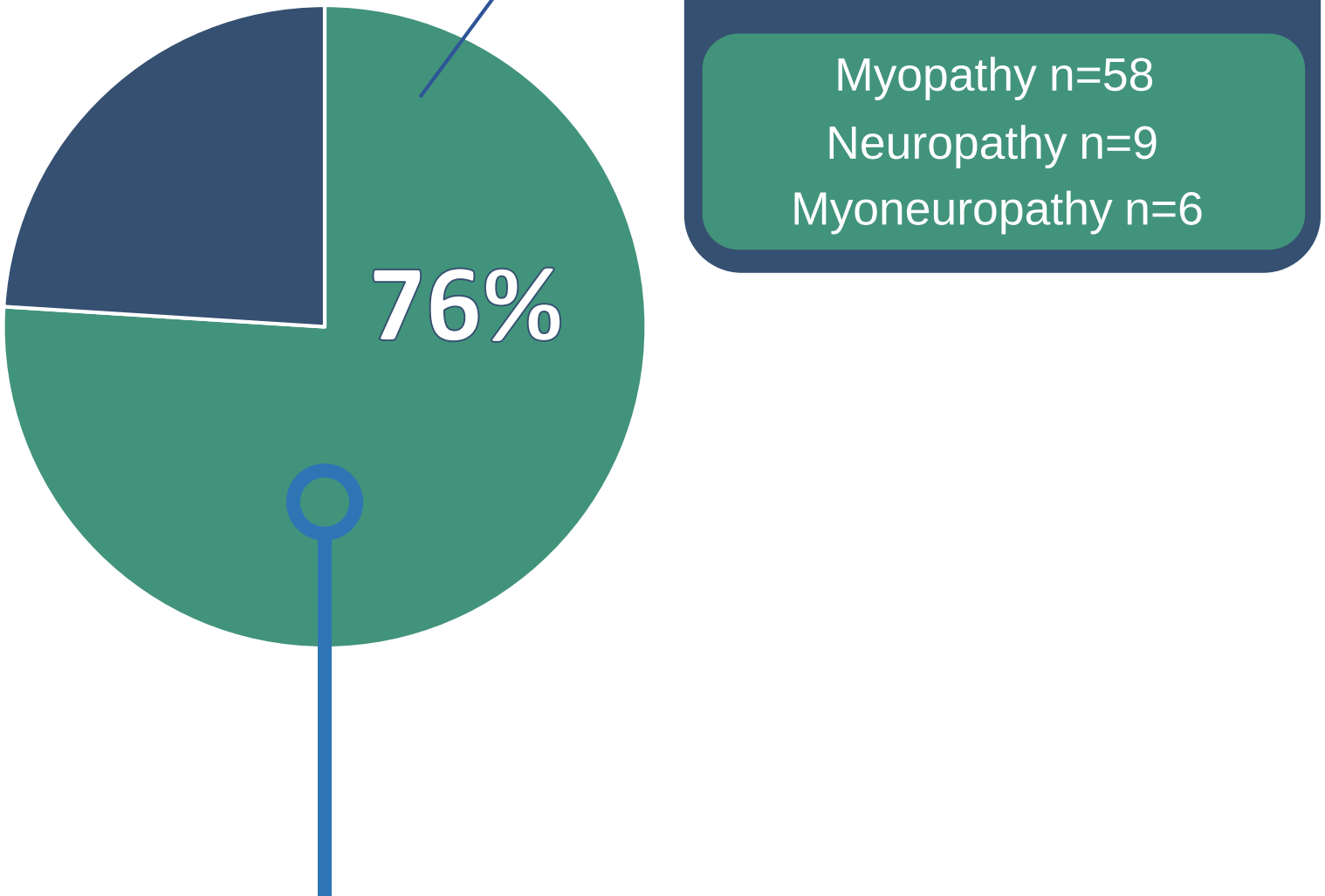
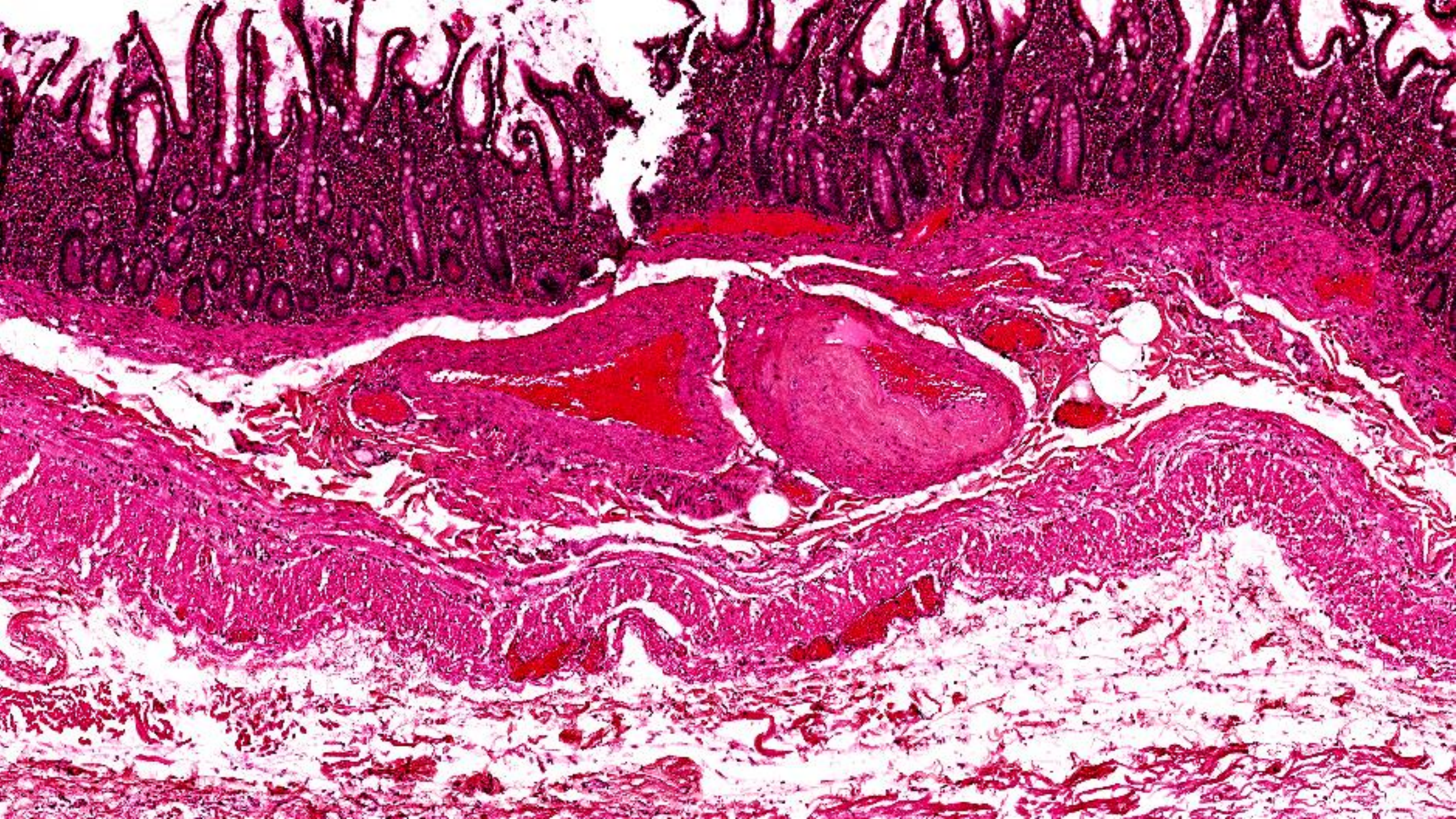
MT-TV n=1
mt-DNA^{DEL} n=2

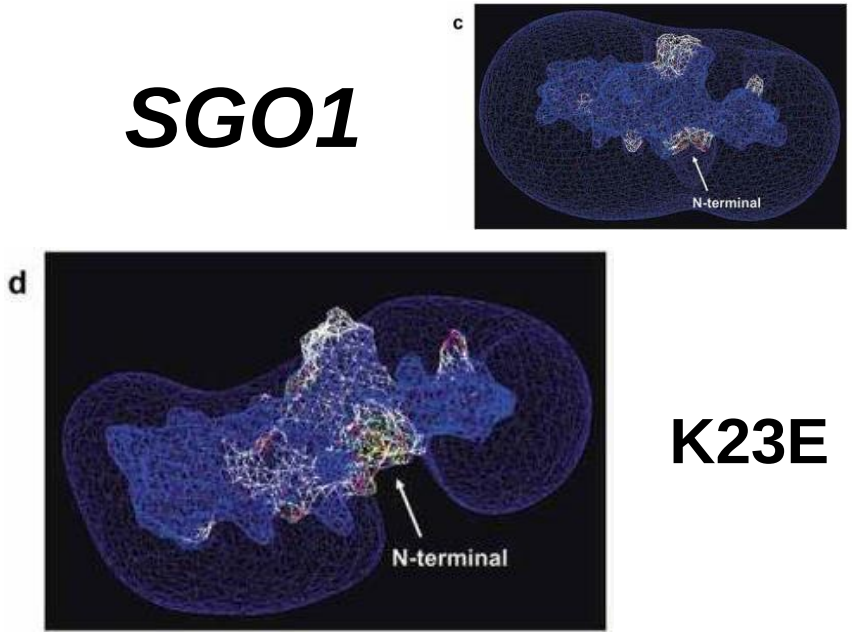
Richards S et al. Genet Med. 2015 May;17(5):405-24.

Analyse histologique, n=96



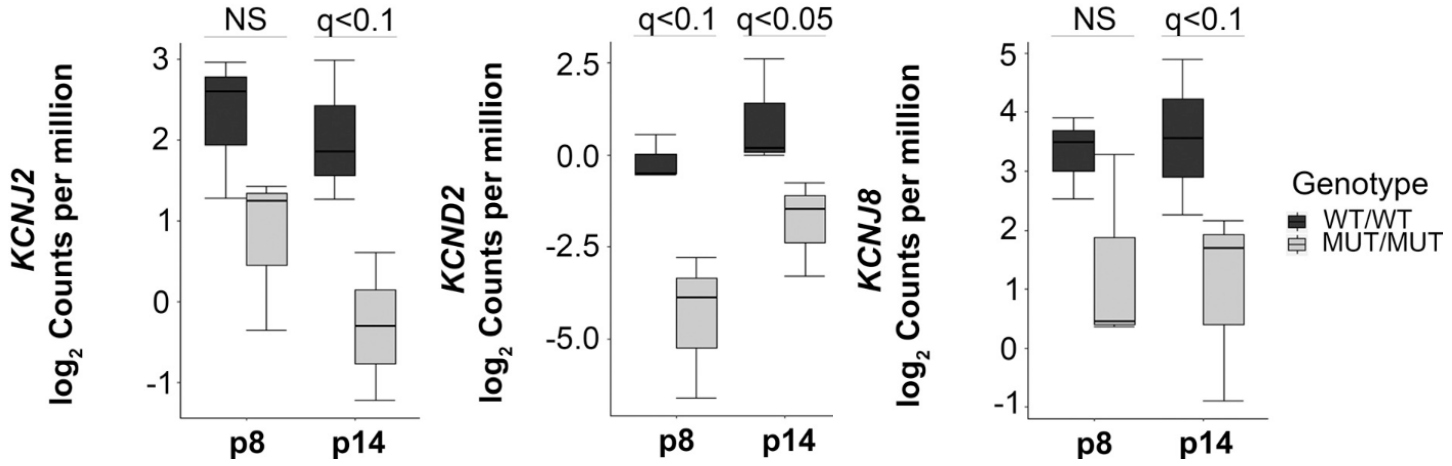


SGO1



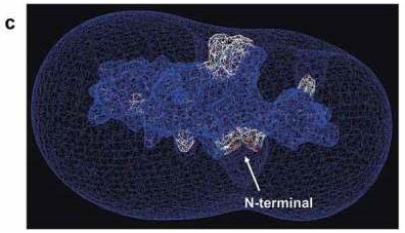
K23E

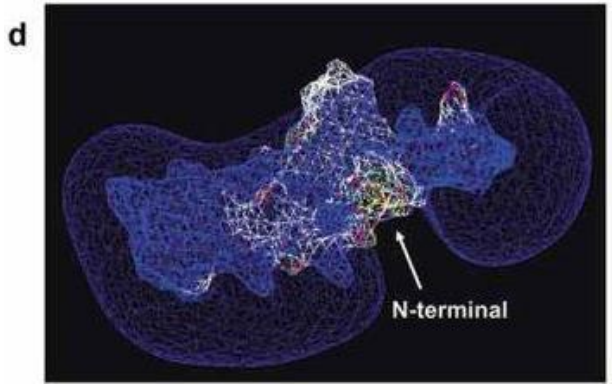
Piché et al. Cell Cycle, 2019;18(21):2828–2848.



Piché et al. Cell Mol Gastroenterol Hepatol, 2019; 7(2):441-431.

SGO1





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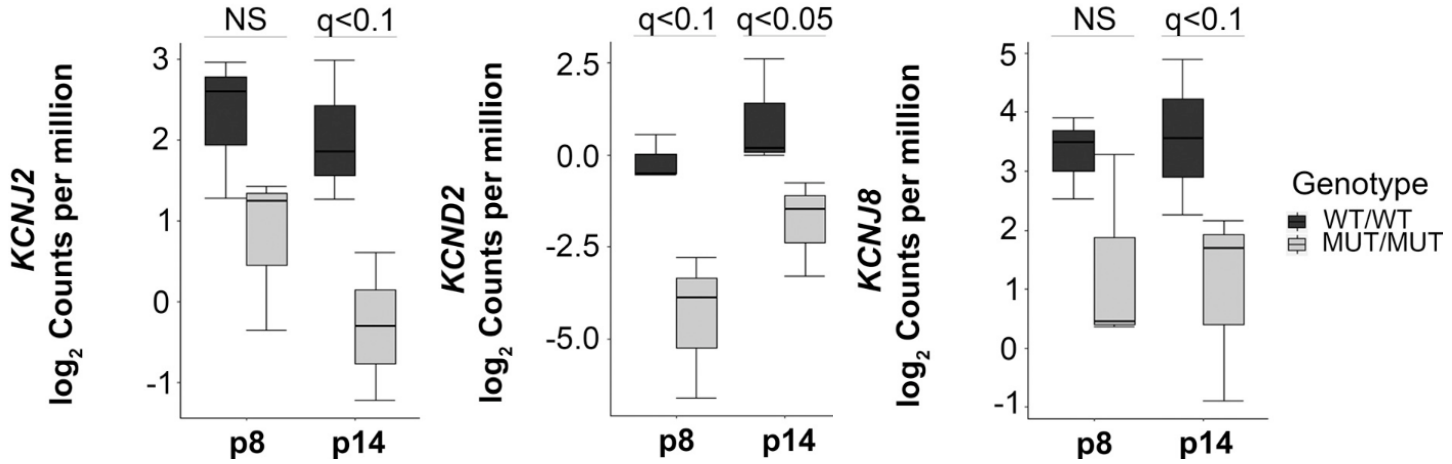

Piché et al. Cell Mol Gastroenterol Hepatol, 2019; 7(2):441-431.

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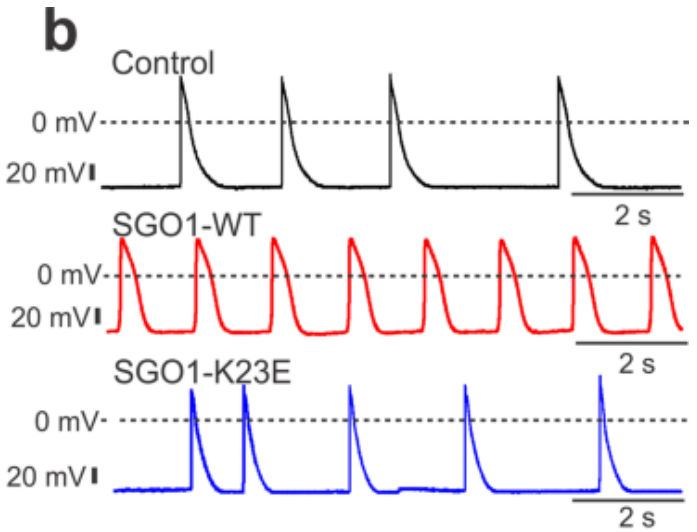


Liu et al. Nat Commun 2021;12, 2551.

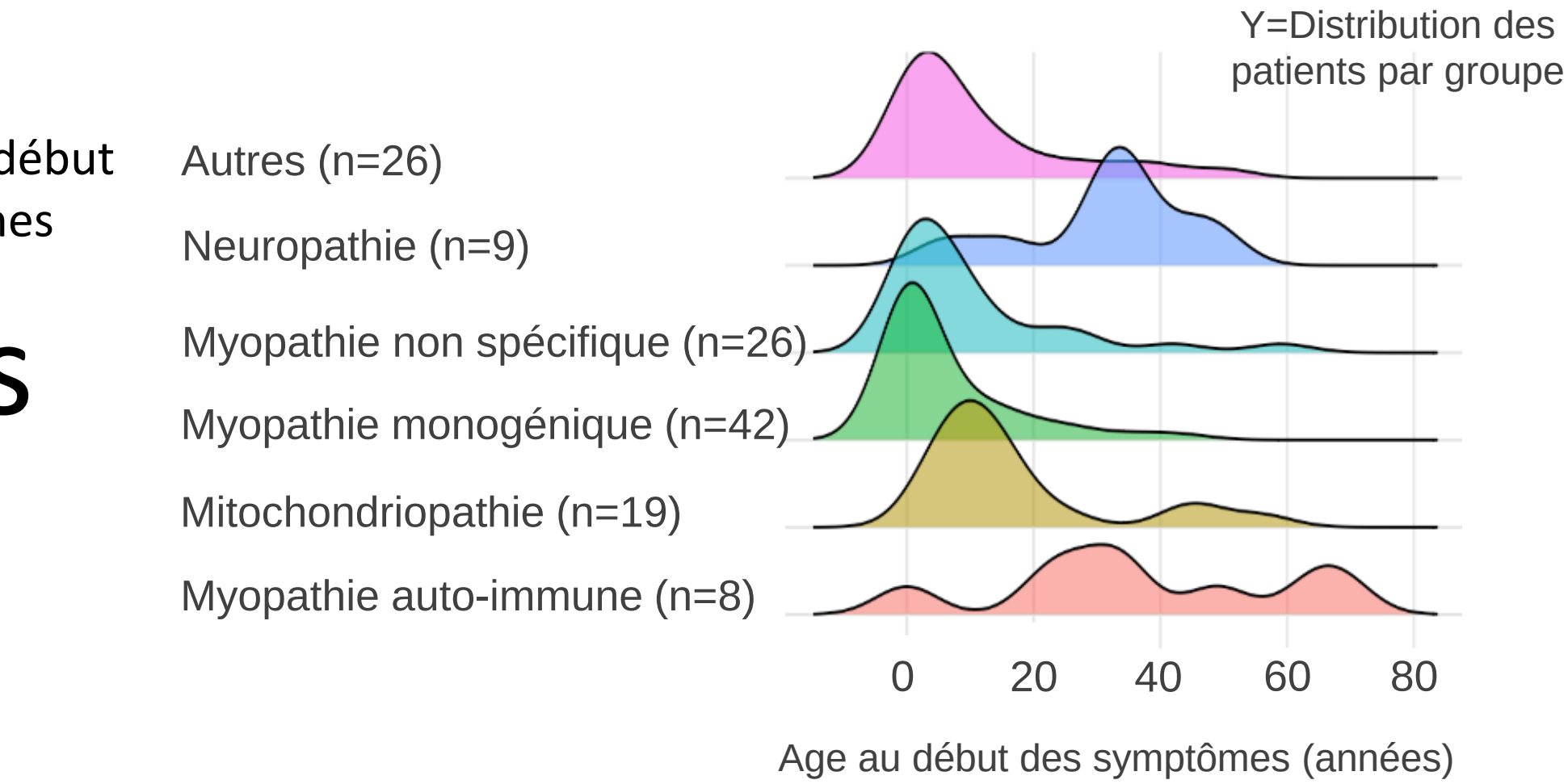
106 patients avec caractérisation (82%)

24 patients idiopathiques (18%)

Patients n=130	Myopathie monogénique n=42	Mitochondriopathie n=19	Myopathie non spécifique n=26	Myopathie auto-immune n=8	Neuropathie n=9	Autres n=26
Histologie n=96	Myopathie n=28 Normal n=4	Myopathie n=3 Mixte n=3	Myopathie n=20 Mixte n=3	Myopathie n=7 Non concluante n=1	Neuropathie n=9	Normal n=9 Non concluante n=9
Genetique n=112	*ACTG2* n=29 *MYH11* n=9 *SGO1* n=2 *FLNA* n=2	*TYMP* n=9 *POLG* n=3 *MT-TL1* n=3 *MT-ATP6* n=1 *MT-TV* n=1 mt-DNA^{DEL} n=2	Negative n=20	*STAT1* n=1 *SOCS1* n=1	Negative n=6	*POGZ* n=1 *TTC37* n=1 Negative n=21

Âge médian au début des symptômes

6 ans



Autres (n=26)

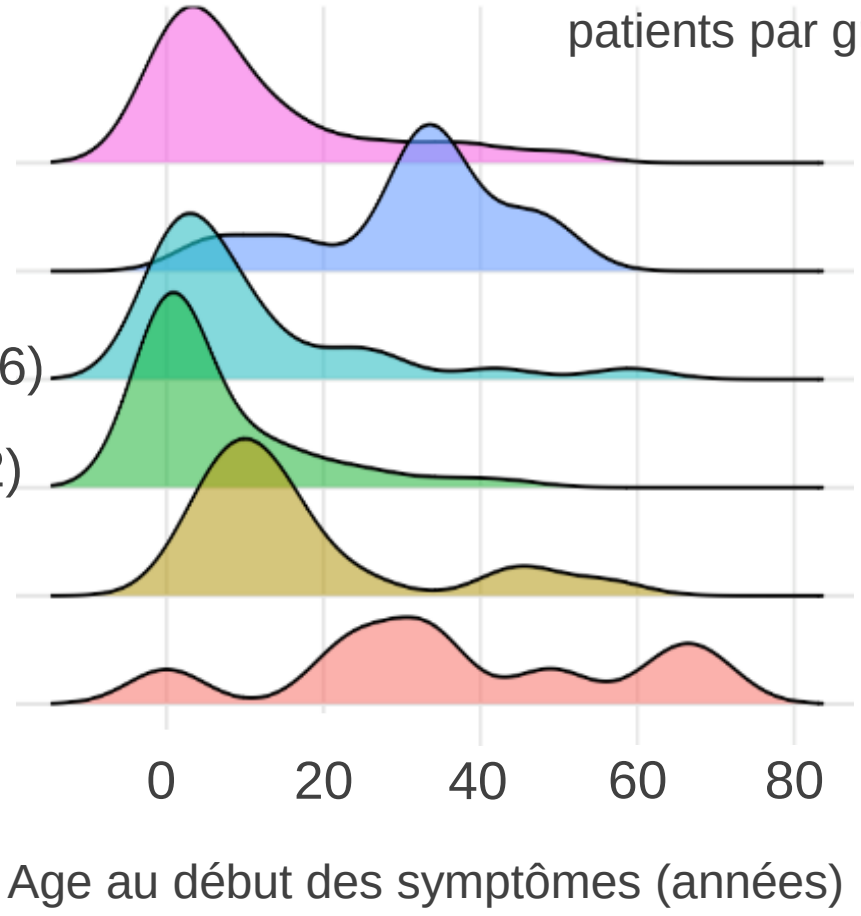
Neuropathie (n=9)

Myopathie non spécifique (n=26)

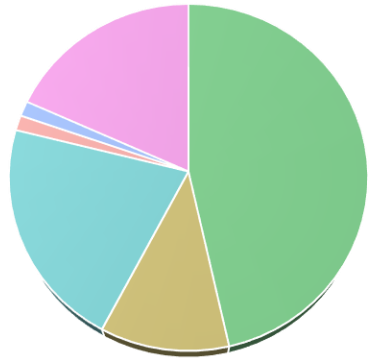
Myopathie monogénique (n=42)

Mitochondriopathie (n=19)

Myopathie auto-immune (n=8)



Admission en pédiatrie
(n=71, 55%)



Admission dans un service d'adulte
(n=59, 45%)

Table 1. Characteristics of Patients According to Histogenetic Features

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	Overall (n=130)	MM (n=42)	Mito (n=19)	UM (n=26)	AI M (n=8)	Neuro (n=9)	Others (n=26)	p-value
Symptoms before diagnosis — no./total no. (%)								
Pyloric stenosis	6 (5)	6 (14)	0 (0)	0 (0)	0 (0)	0 (0)	0 (0)	0.063
Constipation	55/129 (43)	14 (33)	9/18 (50)	12 (46)	1 (13)	4 (44)	15 (58)	0.193
Diarrhoea without constipation	22/129 (17)	2 (5)	3/18 (17)	6 (23)	5 (63)	2 (22)	4 (15)	0.005
Vomiting	63/129 (49)	23 (55)	6/18 (33)	16 (62)	5 (63)	4 (44)	9 (35)	0.242
Volvulus	12 (9)	5 (12)	0 (0)	2 (8)	0 (0)	1 (11)	4 (15)	0.512
Symptoms during the follow-up— no./total no. (%)								
Chronic abdominal pain	70 (54)	17 (41)	10 (53)	15 (58)	5 (63)	5 (56)	18 (69)	0.313
Dysphagia	29/118 (25)	5/40 (13)	7/16 (44)	7/22 (32)	2/8 (25)	5/8 (63)	3/24 (13)	0.009
Volvulus	28/129 (22)	10 (24)	0 (0)	6/25 (23)	2 (25)	2 (22)	8 (31)	0.118
Lower GI bleeding	31/129 (24)	17 (41)	3 (16)	4/25 (16)	2 (25)	0 (0)	5 (19)	0.062
Urinary symptoms	52 (40)	27 (64)	9 (47)	9 (35)	1 (13)	0 (0)	6 (23)	< 0.001
Cardiopathy	39 (30)	14 (33)	3 (16)	12 (46)	3 (38)	1 (11)	6 (23)	0.194
Epilepsy	9 (7)	1 (2)	2 (11)	1 (4)	0 (0)	1 (11)	4 (15)	0.269
Cognitive impairment	7 (5)	1 (2)	6 (32)	0 (0)	0 (0)	0 (0)	0 (0)	< 0.001
Sensorimotor neuropathy	14 (11)	0 (0)	12 (63)	0 (0)	0 (0)	2 (22)	0 (0)	< 0.001
Ear, Nose, Throat (ENT) impairment	9 (7)	0 (0)	6 (32)	1 (4)	0 (0)	1 (11)	1 (4)	0.001
Ophthalmic impairment	9 (7)	0 (0)	8 (42)	0 (0)	0 (0)	0 (0)	1 (4)	< 0.001
Leukoencephalopathy (MRI)	8/80 (10)	0/24 (0)	7/15 (47)	0/14 (0)	0/5 (0)	0/5 (0)	1/17 (6)	< 0.001

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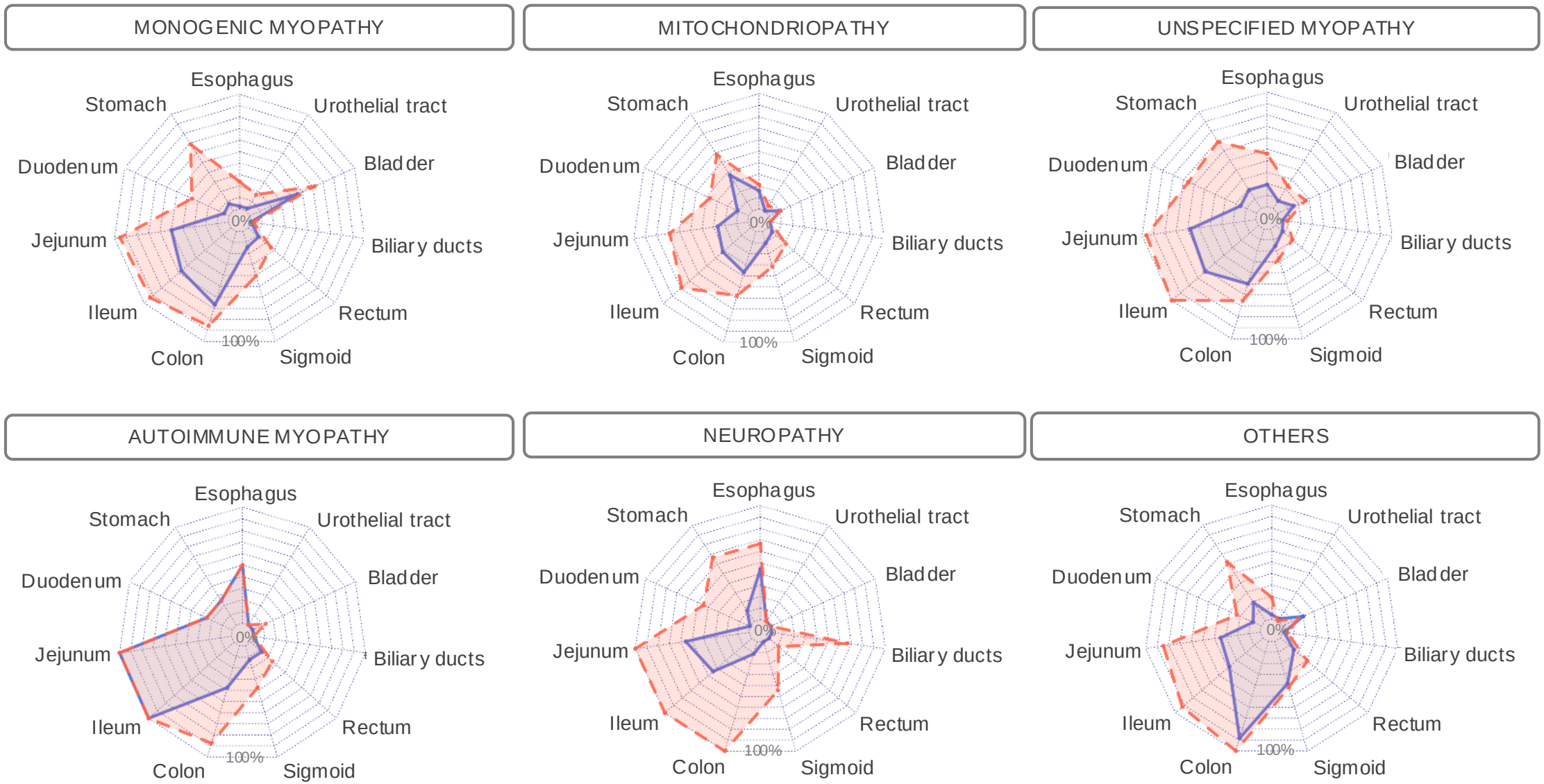
Ear, Nose, Throat (ENT) impairment	9 (7)	0 (0)	6 (32)	1 (4)	0 (0)	1 (11)	1 (4)	0.001
Ophthalmic impairment	9 (7)	0 (0)	8 (42)	0 (0)	0 (0)	0 (0)	1 (4)	< 0.001
Leukoencephalopathy (MRI)	8/80 (10)	0/24 (0)	7/15 (47)	0/14 (0)	0/5 (0)	0/5 (0)	1/17 (6)	< 0.001

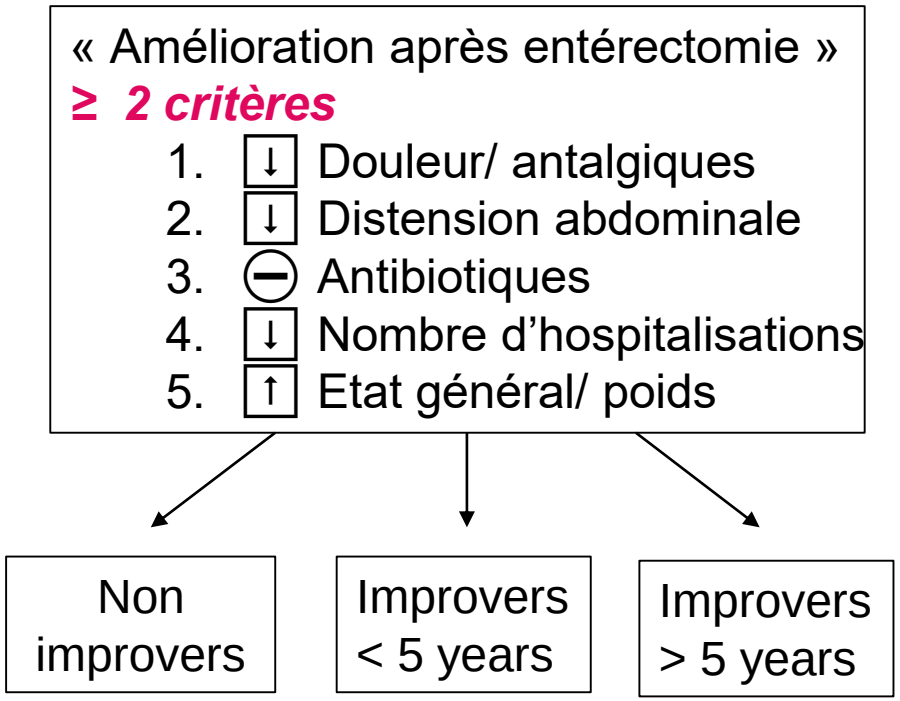
Table 1. Characteristics of Patients According to Histogenetic Features

Abbreviations: MM = Monogenic Myopathy; Mito = Mitochondriopathy; UM = Unspecified Myopathy; AI M = Autoimmune Myopathy; Neuro = Neuropathy.

	Overall (n=130)	MM (n=42)	Mito (n=19)	UM (n=26)	AI M (n=8)	Neuro (n=9)	Others (n=26)	p-value
Symptoms before diagnosis — no./total no. (%)								
Pyloric stenosis	6 (5)	6 (14)	0 (0)	0 (0)	0 (0)	0 (0)	0 (0)	0.063
Constipation	55/129 (43)	14 (33)	9/18 (50)	12 (46)	1 (13)	4 (44)	15 (58)	0.193
Diarrhoea without constipation	22/129 (17)	2 (5)	3/18 (17)	6 (23)	5 (63)	2 (22)	4 (15)	0.005
Vomiting	63/129 (49)	23 (55)	6/18 (33)	16 (62)	5 (63)	4 (44)	9 (35)	0.242
Volvulus	12 (9)	5 (12)	0 (0)	2 (8)	0 (0)	1 (11)	4 (15)	0.512
Symptoms during the follow-up— no./total no. (%)								
Chronic abdominal pain	70 (54)	17 (41)	10 (53)	15 (58)	5 (63)	5 (56)	18 (69)	0.313
Dysphagia	29/118 (25)	5/40 (13)	7/16 (44)	7/22 (32)	2/8 (25)	5/8 (63)	3/24 (13)	0.009
Volvulus	28/129 (22)	10 (24)	0 (0)	6/25 (23)	2 (25)	2 (22)	8 (31)	0.118
Lower GI bleeding	31/129 (24)	17 (41)	3 (16)	4/25 (16)	2 (25)	0 (0)	5 (19)	0.062
Urinary symptoms	52 (40)	27 (64)	9 (47)	9 (35)	1 (13)	0 (0)	6 (23)	< 0.001
Cardiopathy	39 (30)	14 (33)	3 (16)	12 (46)	3 (38)	1 (11)	6 (23)	0.194
Epilepsy	9 (7)	1 (2)	2 (11)	1 (4)	0 (0)	1 (11)	4 (15)	0.269
Cognitive impairment	7 (5)	1 (2)	6 (32)	0 (0)	0 (0)	0 (0)	0 (0)	< 0.001
Sensorimotor neuropathy	14 (11)	0 (0)	12 (63)	0 (0)	0 (0)	2 (22)	0 (0)	< 0.001
Ear, Nose, Throat (ENT) impairment	9 (7)	0 (0)	6 (32)	1 (4)	0 (0)	1 (11)	1 (4)	0.001
Ophthalmic impairment	9 (7)	0 (0)	8 (42)	0 (0)	0 (0)	0 (0)	1 (4)	< 0.001
Leukoencephalopathy (MRI)	8/80 (10)	0/24 (0)	7/15 (47)	0/14 (0)	0/5 (0)	0/5 (0)	1/17 (6)	< 0.001

2A Dilation observed on initial imaging (% of patients) — Dilation observed on last imaging (% of patients)




« Amélioration après entérectomie »
≥ 2 critères
1. ↓ Douleur/ antalgiques
2. ↓ Distension abdominale
3. ⊖ Antibiotiques
4. ↓ Nombre d'hospitalisations
5. ↑ Etat général/ poids
Non improvers
Improvers < 5 years
Improvers > 5 years

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Non improvers	Improvers < 5 years	Improvers > 5 years

2D

Survival

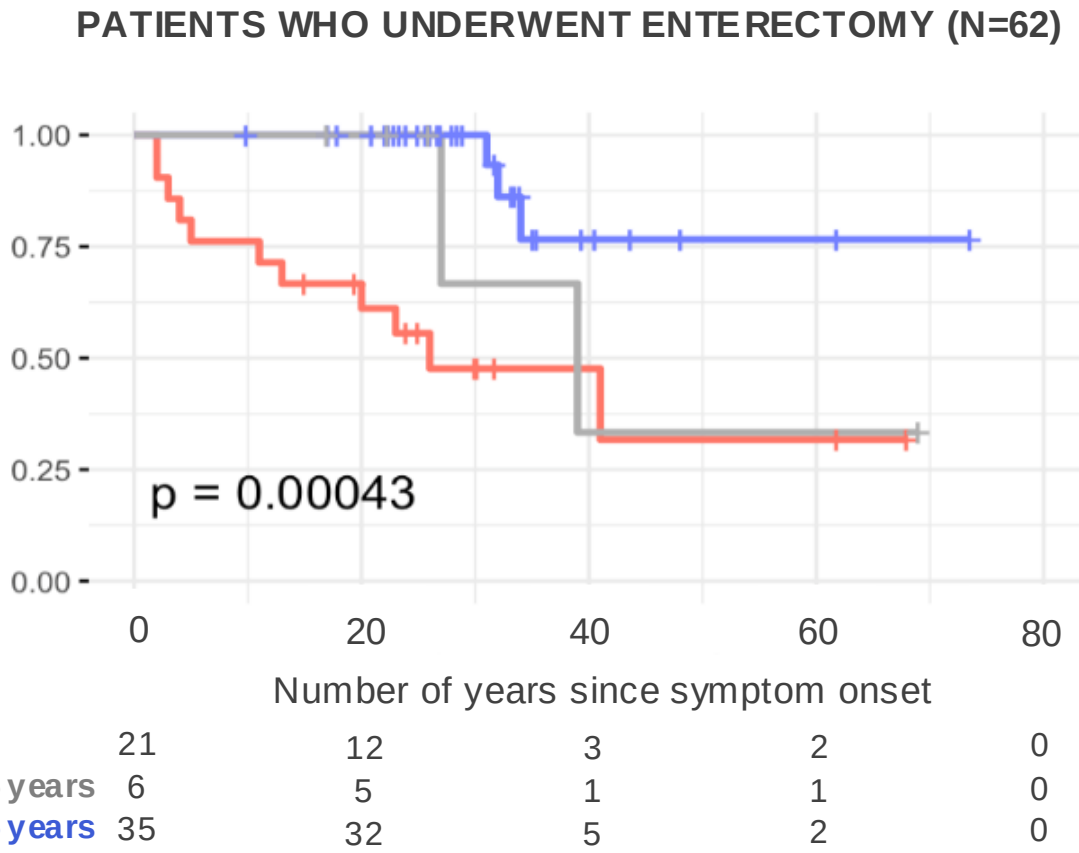

Table 2. Patients' Outcomes in the Six Histogenetic Groups.

Abbreviations: MM = Monogenic Myopathy; Mito = Mitochondriopathy; UM = Unspecified Myopathy; AI M = Autoimmune Myopathy; Neuro = Neuropathy.

	Overall (n=130)	MM (n=42)	Mito (n=19)	UM (n=26)	AI M (n=8)	Neuro (n=9)	Others (n=26)	p-value
Improvement after surgery — no./total no. (%)								
Ileostomy	42/55 (76)	21/24 (88)	3/3 (100)	9/16 (56)	1/3 (33)	3/4 (75)	5/5 (100)	0.054
Jejunostomy	17/29 (59)	7/9 (78)	2/2 (100)	5/10 (50)	0/3 (0)	2/2 (100)	1/3 (33)	0.086
Small bowel resection	41/62 (66)	22/23 (96)	2/5 (40)	8/14 (57)	0/6 (0)	5/6 (83)	4/8 (50)	< 0.001
Colon resection	38/64 (59)	20/22 (91)	2/5 (40)	7/12 (58)	0/3 (0)	1/7 (17)	8/16 (50)	< 0.001
5-year survival after multivisceral transplantation (%)	3/5 (60)	2/2 (100)	-	1/3 (33)	-	-	-	-
Death (%)	25 (19)	2 (5)	6 (32)	6 (23)	7 (88)	2 (22)	2 (8)	< 0.001
Age at death (years, mean, range)	42 (21-82)	35 (30-40)	41 (29-59)	35 (21-70)	45 (31-82)	55 (40-71)	47 (33-60)	0.752

odds ratio: 19.77, 95% CI: 3.55–504.66; p<0.001

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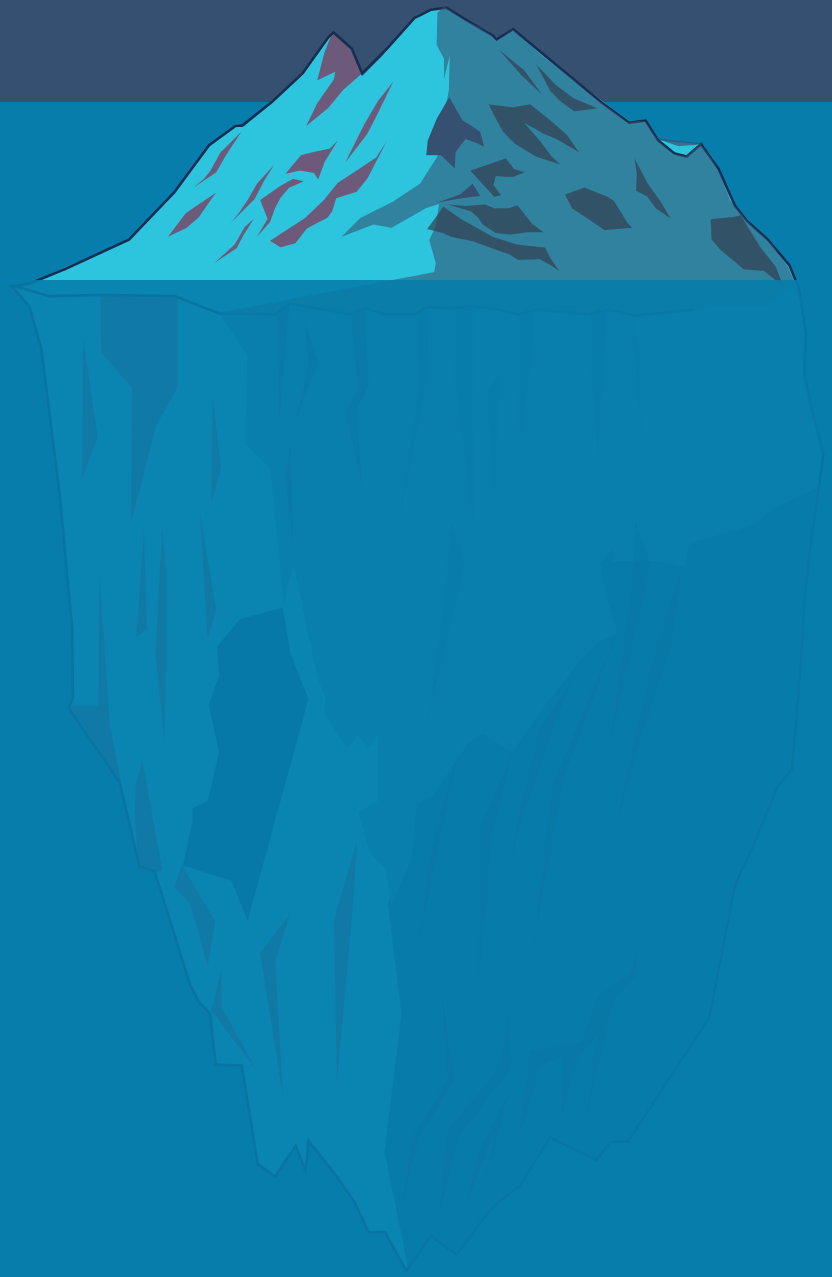

58%

58%

82%

Patients avec une analyse histologique (n=96)

Survie

1.00

0.75

0.50

0.25

0.00

Musculeuse non atrophique

Musculeuse atrophique

0

20

40

60

80

Nombre d’années depuis le début des symptômes

DDW
Digestive Disease Week®

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47th ESPEN Congress
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13-16 September 2025
Prague Congress Center

Prof. Francisca Joly
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HAVE A NICE DAY