Distribution and Temporal Progression of Aortopathy in Genetically Confirmed Marfan Syndrome

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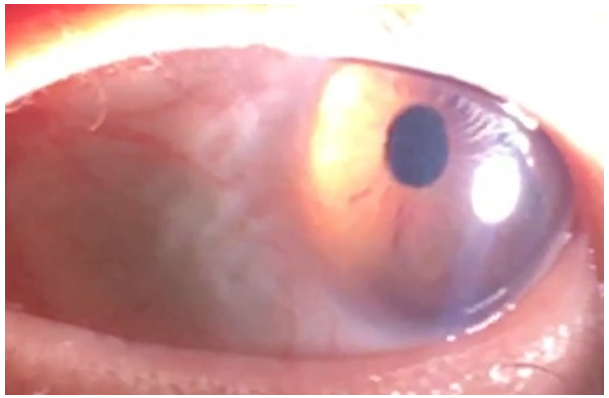
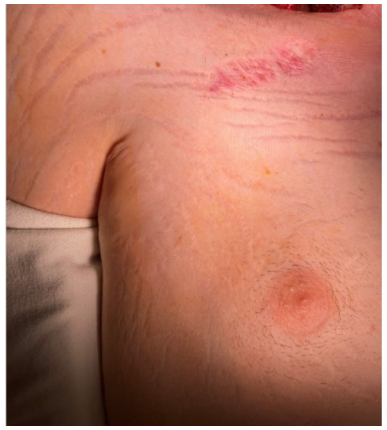
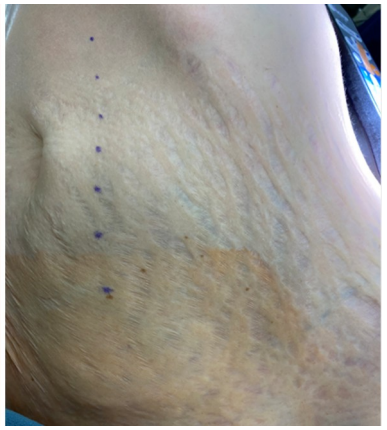
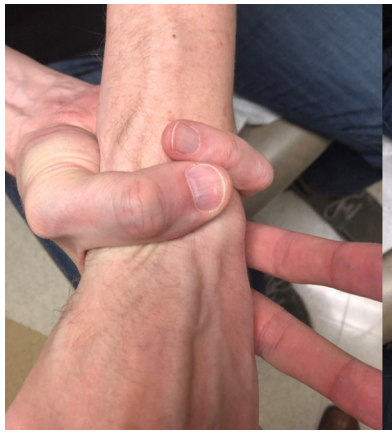
Marfan Syndrome

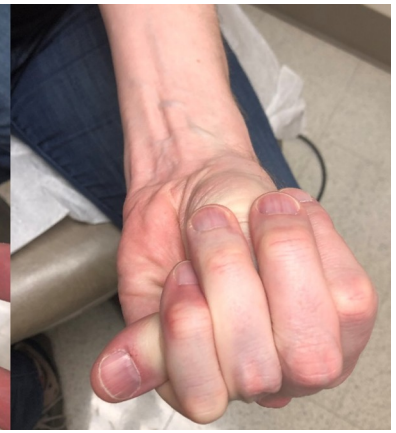
Connective tissue disorder caused by pathogenic variants in *FBN1* → encodes fibrillin-1, an important component of microfibrils in the extracellular matrix.

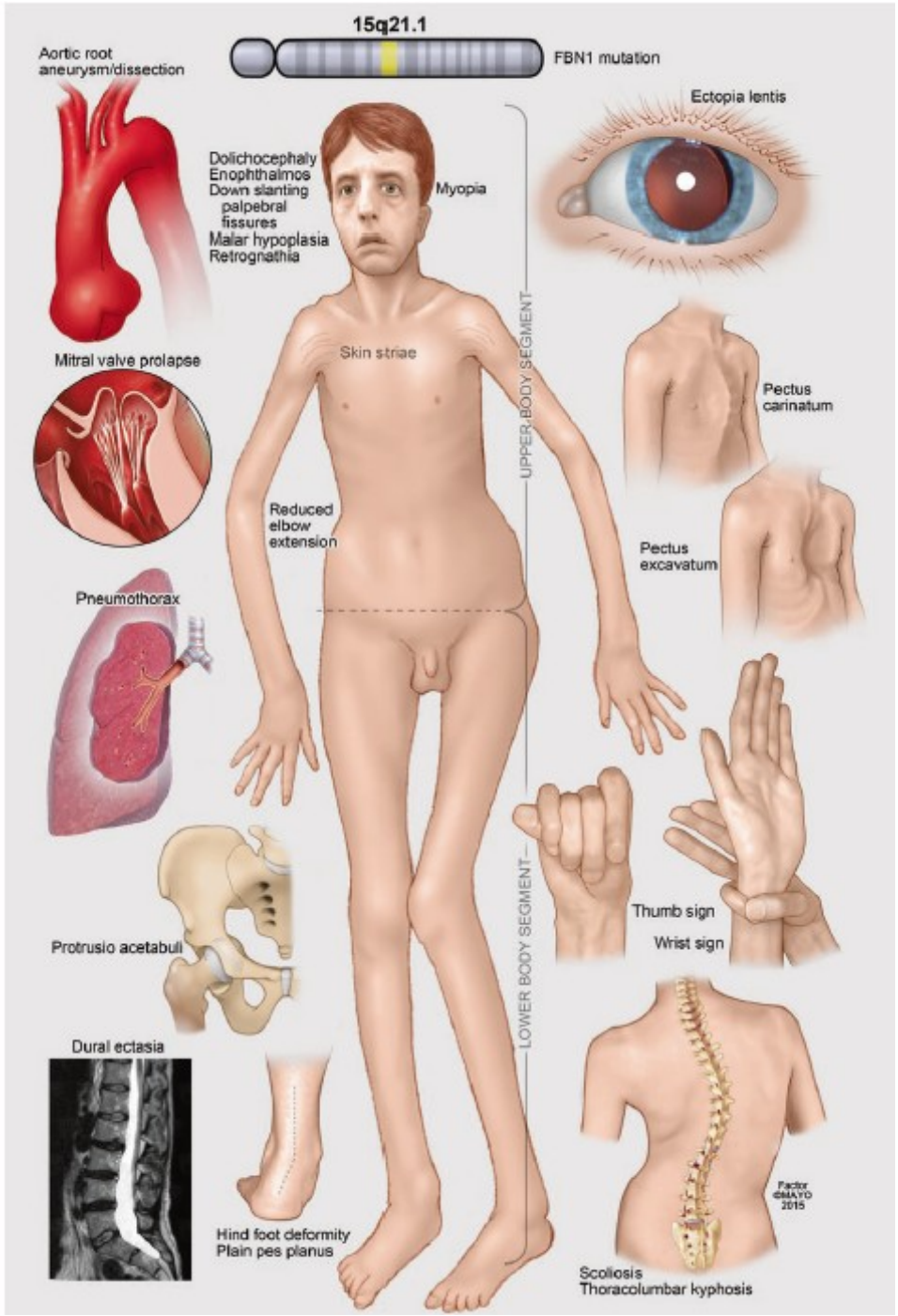












2022 ACC/AHA Guidelines

- Most of the studies describe patients with clinical diagnosis of MFS as genetic testing was not widely available

KNOWLEDGE GAP:
What is the natural history beyond the aortic root in genetically confirmed MFS?

Study aim

To evaluate distribution and temporal progression of aortopathy in patients with MFS, using age at diagnosis and aortic zone-based phenotyping.

Methods

A single-center retrospective review of all patients with ICD-10-CM diagnosis code of Q87.4 in 2023-2024

Inclusion: *FBN-1* pathogenic/likely pathogenic variant

Exclusion:

VUS in *FBN1*	No genetic testing	Other genetic aortopathy	No medical records

Ascertainment of aortopathy

Root/Ascending ≥ 4.5 cm
Descending ≥ 4 cm
Abdominal ≥ 3 cm
Common iliac artery ≥ 2 cm

Aortic root dilation: Y/N
Aortic dissection: type A, type B
Age of aneurysm diagnosis
Age of treatment
Intervention modality
Timing of intervention

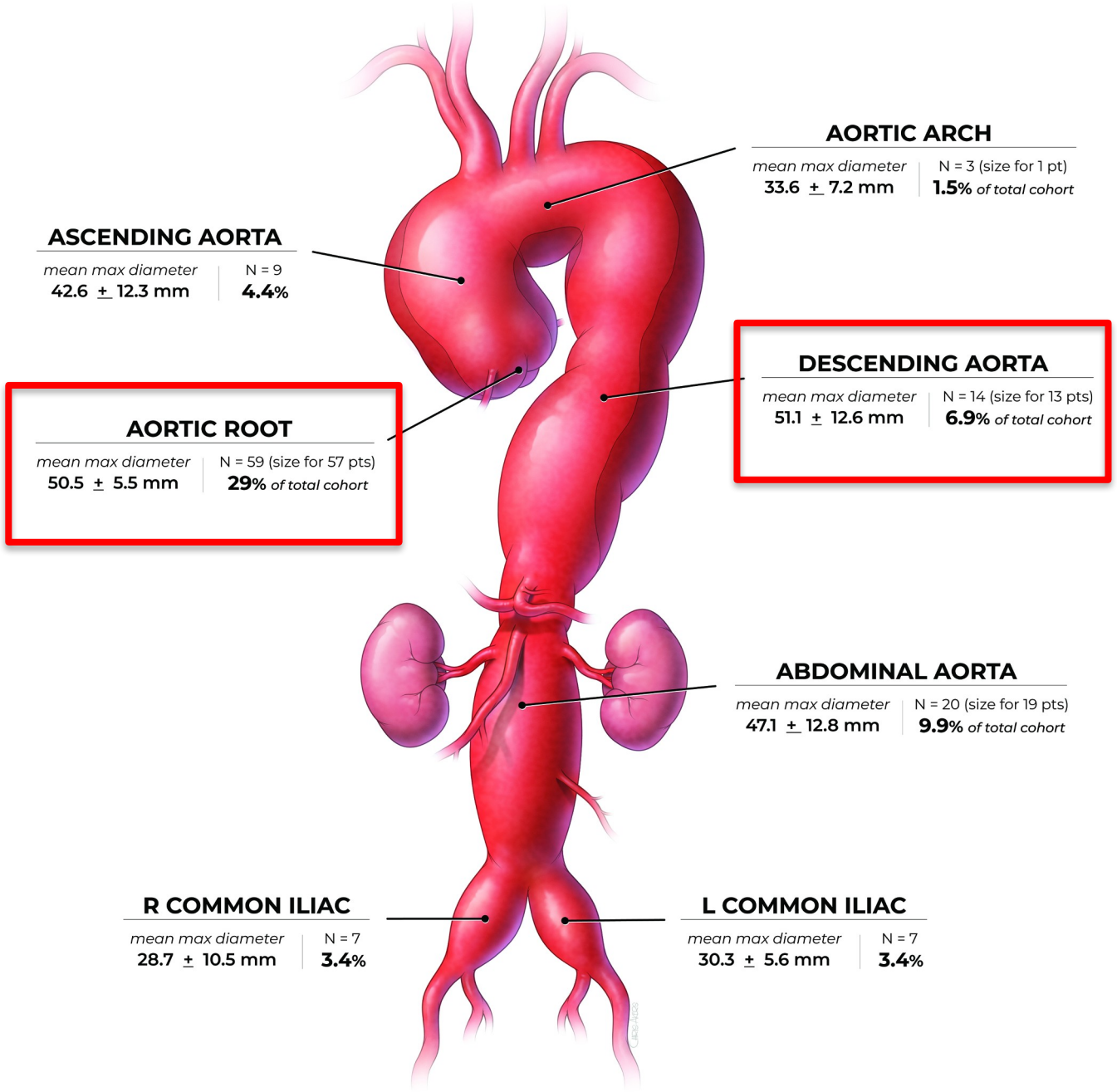
OHSU

1822 patients with diagnostic codes for MFS
203 patients with P/LP *FBN1* variants

	Cohort (N=203)
Current age	28 ± 17.7
Age of genetic testing	19.7 ± 17.6
Female	52.2%
Pediatric patients	35%

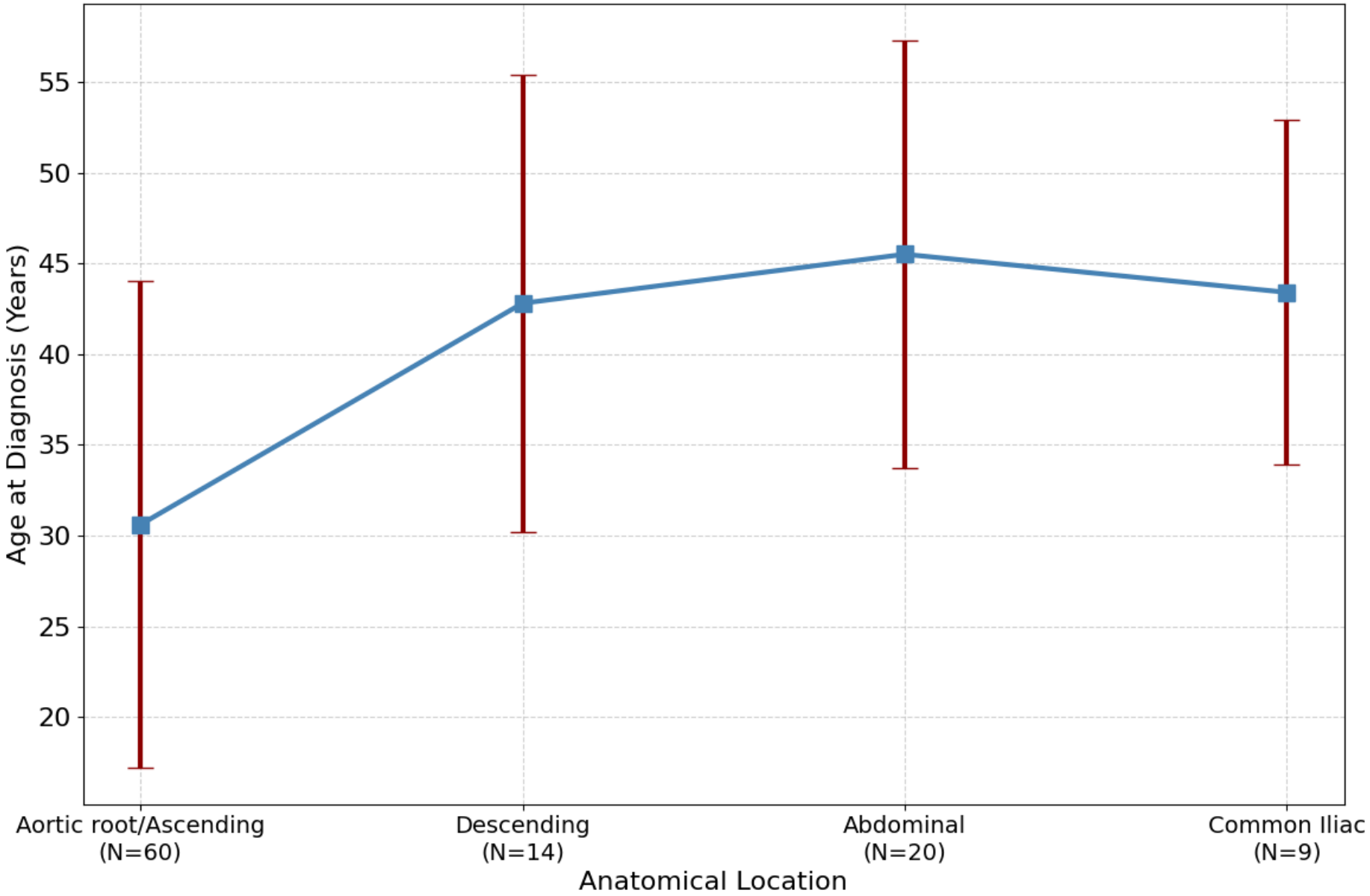
135/203 (66.5%) had an aortopathy
64/203 (31.5%) had aortic

	Cohort (N=203)	Mean age
Aortic aneurysm	31.5%	31.9 ± 14.4
Aortic root/Ascending aortic aneurysm	29.5%	30.6 ± 13.4
Descending aneurysm	6.9%	42.8 ± 12.6
Abdominal aortic aneurysm	9.9%	45.5 ± 11.8
Common Iliac aneurysm	4.4%	43.4 ± 9.5
Type A dissection	5.4%	32.6 ± 5.4
Type B dissection	6.4%	41.5 ± 11.2

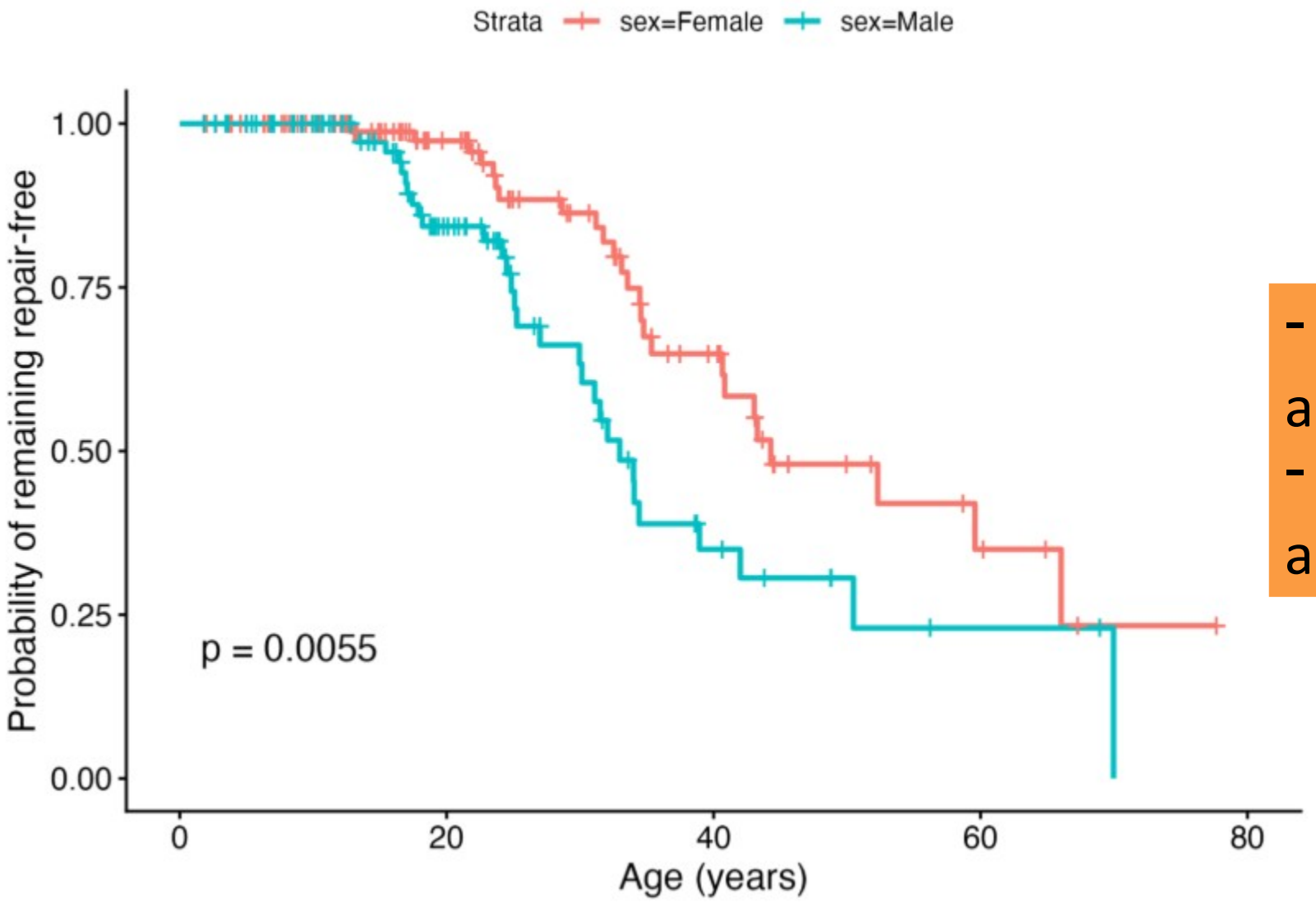


Zone 0 aorthopathy was the most common

Cranio-Caudal Disease Progression by age

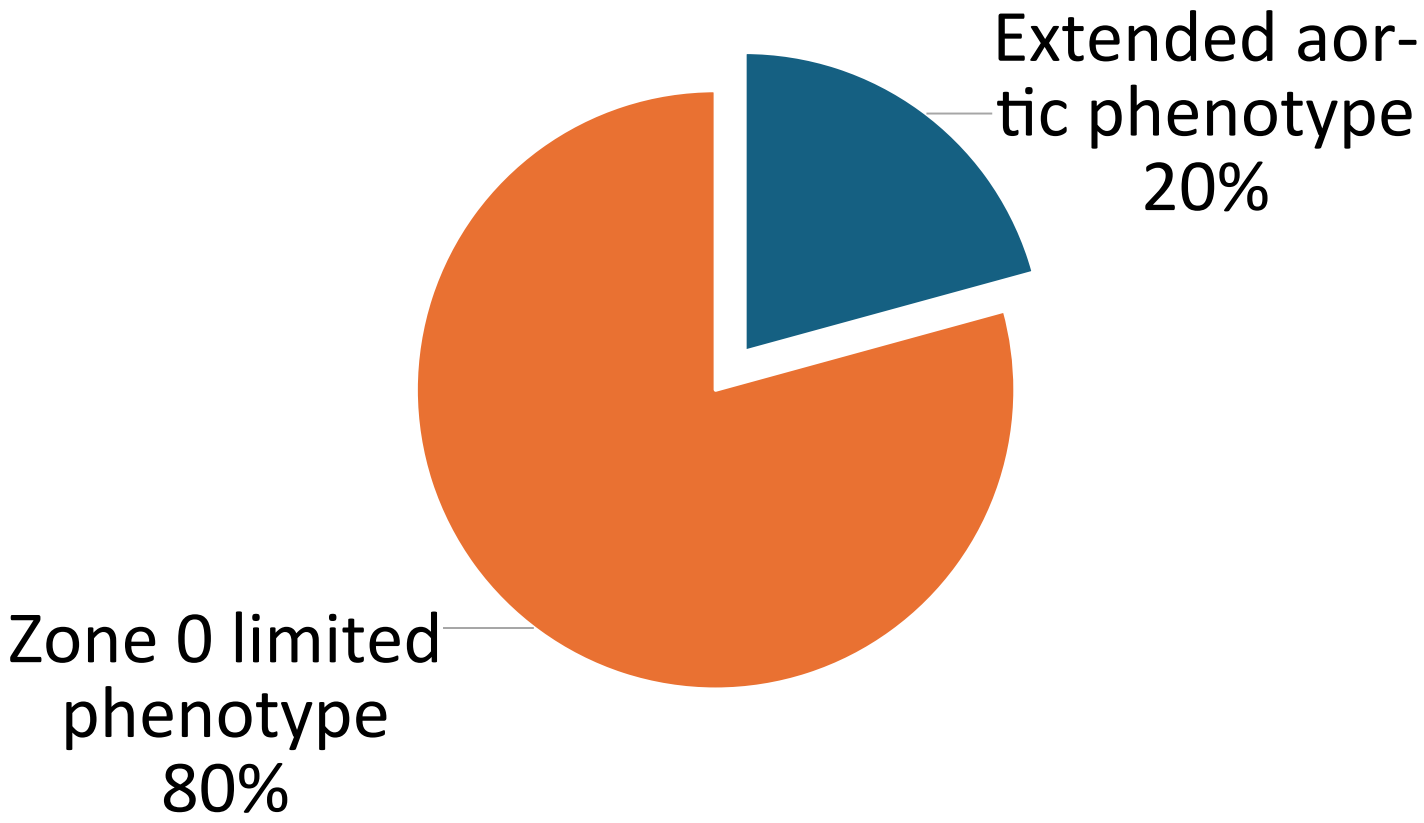


Men were more likely to have any aortic repair at younger age



- Men had 30 distinct aortic operations
- Women had 24 distinct aortic operations

Aortic Phenotype



- 81.5% patients with extended aortic phenotype had aortic dissection (11 TAAD, 13 TBAD)
- 6 had distal aortic aneurysms without dissection

- 25/27 (92.6%) patients with extended aortic phenotype had 45 operations
- 30/108 (27.8%) patients with zone 0 limited phenotype had 31 operations

Aortic phenotype associated features

	Zone 0 limited aortic phenotype (N=108)	Extended aortic Phenotype (N=27)	p- value
Average age at the time of study	24.3	50.6	<.001
Average age at genetic dx	16.4	43.9	<.001
Pediatric genetic dx	63	7.4%	<.001
Hypertension	17.6%	81.5%	<.001
Obstructive sleep apnea	10.2%	44.4%	<.001
Migraine headaches	15.7%	37%	.027
Osteoarthritis	7.4%	40.7%	<.001
Hernias	16.7%	44.4%	.004
Dural ectasia	17.6%	40.7%	.020
Tarlov cysts	9.3%	44.4%	<.001

Extended aortic phenotype risk factors

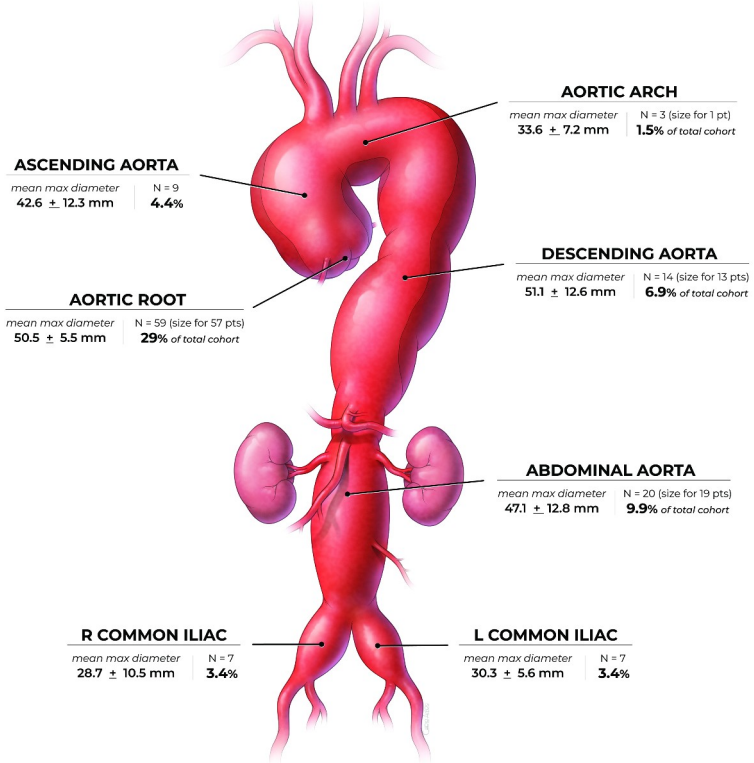
outcome <chr>	model <chr>	OR <dbl>	CI <chr>	p.value <chr>
htn	Logistic (age-adj...	4.7228238	1.40-17.62	0.015
sleep_apnea	Logistic (age-adj...	3.3640647	1.01-11.65	0.050
migraine	Logistic (age-adj...	1.1750699	0.36-3.74	0.786
osteoarthritis	Logistic (age-adj...	1.1725187	0.28-4.55	0.821
joint_hyperextens...	Logistic (age-adj...	0.3057419	0.06-1.16	0.104
hernia	Logistic (age-adj...	1.4385791	0.46-4.42	0.527
dural_ectasia	Logistic (age-adj...	1.3115346	0.41-4.05	0.639
tarlov	Logistic (age-adj...	2.7465254	0.81-9.43	0.103

Variable <chr>	OR <dbl>	CI_low <dbl>	CI_high <dbl>	p-value <chr>
Intercept	0.01	0.00	0.04	<0.001
Age at genetic dia...	1.09	1.04	1.14	<0.001
Hypertension	5.04	1.43	19.22	0.013

- Age of Genetic diagnosis - 9% increase risk
- Hypertension - 5-fold increase risk
- Sleep apnea – high correlation

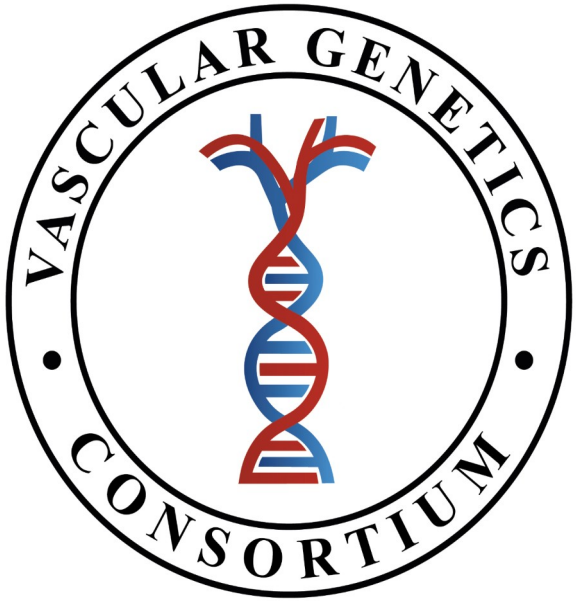
Conclusions

- Aortopathy in MFS progresses in a cranio-caudal pattern.
- Most of the extended aortic phenotype is associated with aortic dissection in individuals without established MFS diagnosis at the time of dissection
- Risk factors: age of genetic diagnosis, hypertensio sleep apnea
- Tarlov cysts appear to be a marker of extended aortic phenotype



Study Limitations and future directions

- Single institution study (next step is multi-center collaboration via vascular genetic consortium)
- Genetic confirmation of MFS is mostly in young individuals
- Next steps: Genotype-phenotype correlation assessment



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