VŠEOBECNÁ FAKULTNÍ NEMOCNICE V PRAZE

1. LÉKAŘSKÁ FAKULTA Univerzita Karlova

SAM A MITRÁLNÍ REGURGITACE:
PATOFYZIOLOGICKÉ ODLIŠNOSTI U FABRYHO CHOROBY A HYPERTROFICKÉ KARDIOMYOPATIE

ALENA VEČEŘOVÁ

II. interní klinika kardiologie a angiologie
Všeobecná fakultní nemocnice
a 1. lékařská fakulta Univerzity Karlovy

12. květen 2026, Praha

XXXIV. Výroční sjezd České kardiologické společnosti

VŠEOBECNÁ FAKULTNÍ NEMOCNICE V PRAZE | 1. LÉKAŘSKÁ FAKULTA Univerzita Karlova

Burden of Valvular Heart Disease in Patients with Fabry Disease

Haran Yogasundaram, MD, MSc · Anish Nikhanj, BSc · Safia Chatur, MD · ... · Nowell M. Fine, MD, SM · John L. Jefferies, MD, MPH · Gavin Y. Oudit, MD, PhD ... Show more

Affiliations & Notes Article Info

	Age, y					
Study	18-44	45-54	55-64	65-74	≧75	Overall
General population						
Nkomo *et al.* (2006)[5] (*n* = 11,911)	0.7% (31/4,351)	0.4% (3/696)	1.9% (23/1,240)	8.5% (328/3,879)	13.2% (230/1,745)	5.2% (615/11,911)
d'Arcy *et al.* (2016)[6] (*n* = 2,500)		NA		3.3% (54/1,621)	11.9% (105/879)	6.0% (159/2,659)
FD						
Kampmann *et al.* (2002)[1] (*n* = 55; 100% women)			NA			0% (0/55)
Weidemann *et al.* (2009)[2] (*n* = 111; 54% women)			NA			2.7% (3/111)
Linhart *et al.* (2000)[3] (*n* = 30; 57% women)			NA			20.0% (6/30)*
Present study	0% (0/46)	12.5% (3/24)	17.6% (3/17)	16.7% (2/12)	100% (2/2)	10.0% (10/100)

Yogasundaram H, Nikhanj A, Chatur S, Qi A, Hagen L, Bailey L, Khan A, Hopkin RJ, Fine NM, Jefferies JL, Oudit GY. Burden of Valvular Heart Disease in Patients with Fabry Disease. J Am Soc Echocardiogr. 2022 Feb;35(2):236-238. doi: 10.1016/j.echo.2021.09.013. Epub 2021 Oct 7. PMID: 34627970.

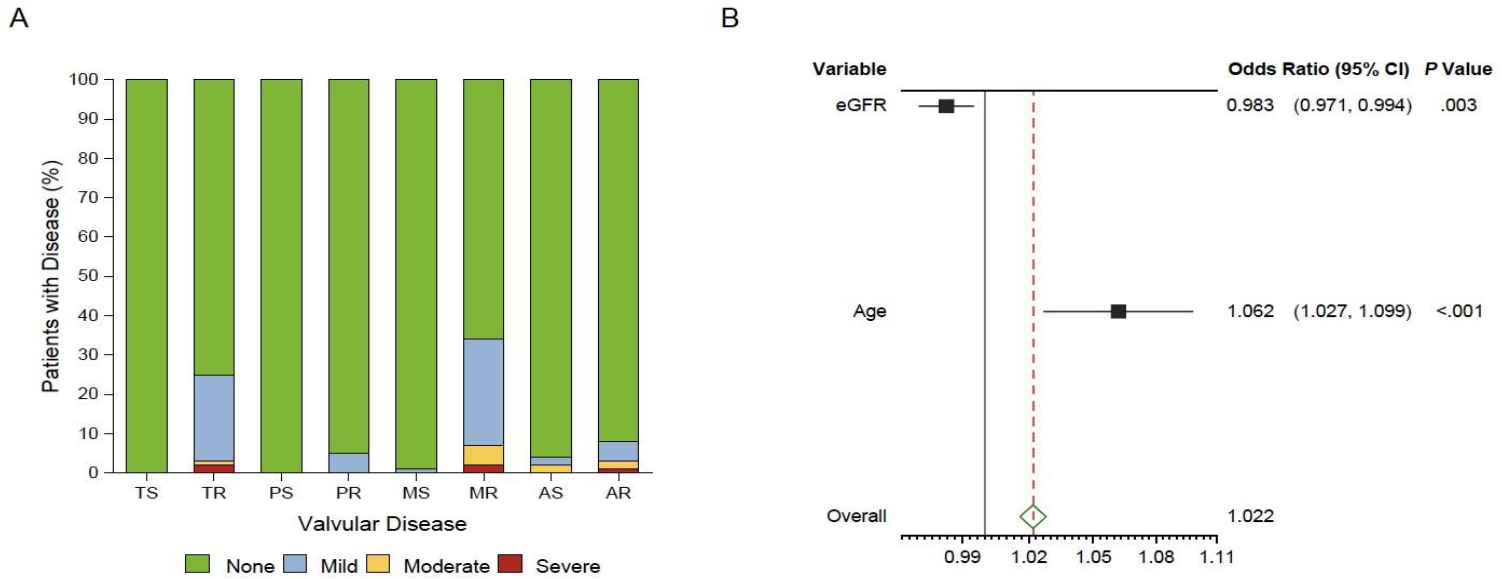
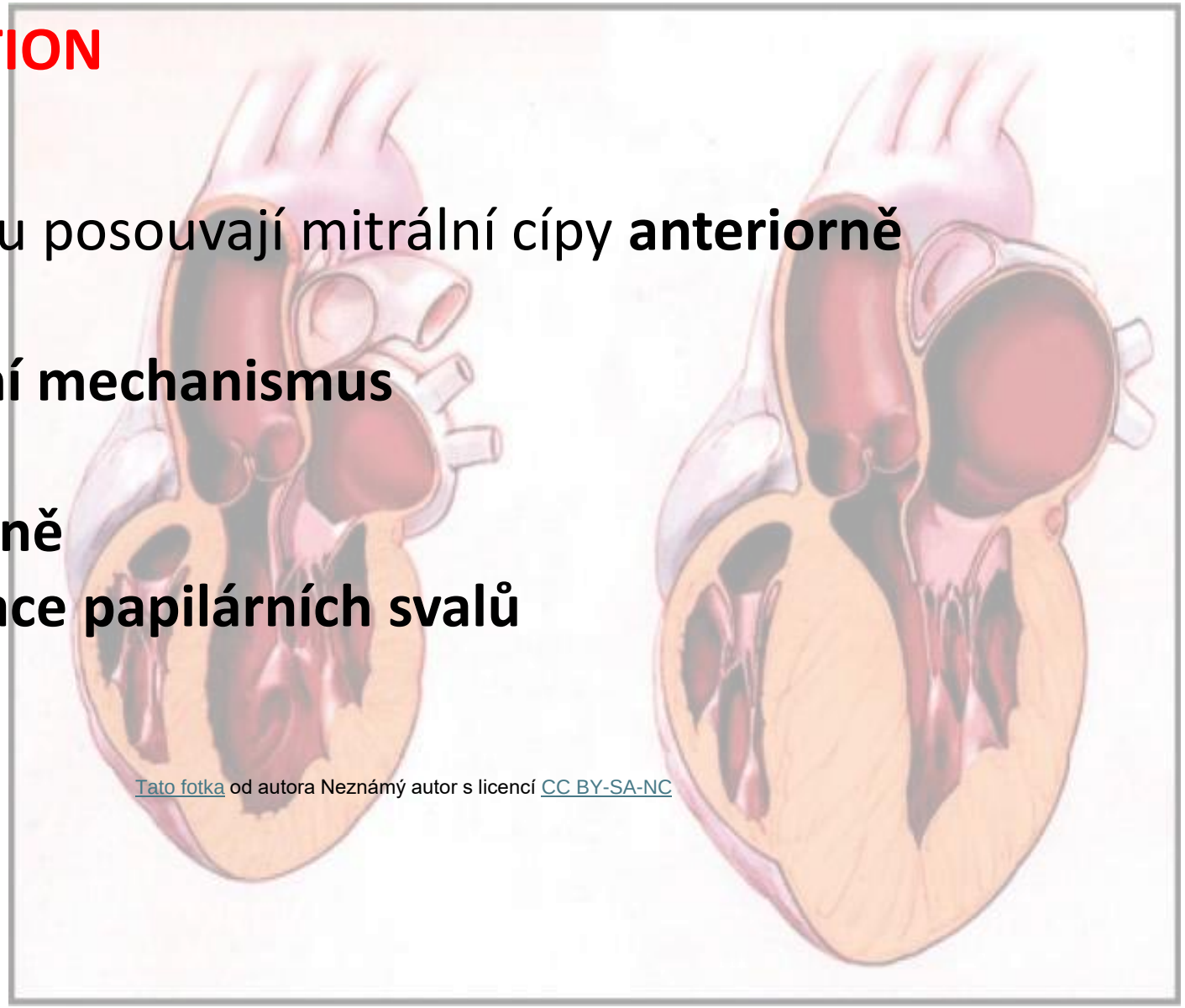


Figure 1 **(A)** Prevalence of VHD in patients with FD. Five patients initially classified with mild to moderate MR were reanalyzed and reclassified as mild (two patients) or moderate (three patients), one instance of mild to moderate aortic stenosis (AS) was downgraded to mild, and one instance of moderate to severe aortic regurgitation (AR) was upgraded to severe. One patient with a bicuspid aortic valve was excluded from the study. Valve morphology is indicated in Supplemental Table 2. **(B)** Forest plot of odds ratios for worse VHD category. Increasing age and worsening estimated glomerular filtration rate (eGFR) were associated with increased odds of VHD. *MR*, Mitral regurgitation; *MS*, mitral stenosis; *PR*, pulmonic stenosis; *PS*, pulmonic stenosis; *TR*, tricuspid regurgitation; *TS*, tricuspid stenosis.

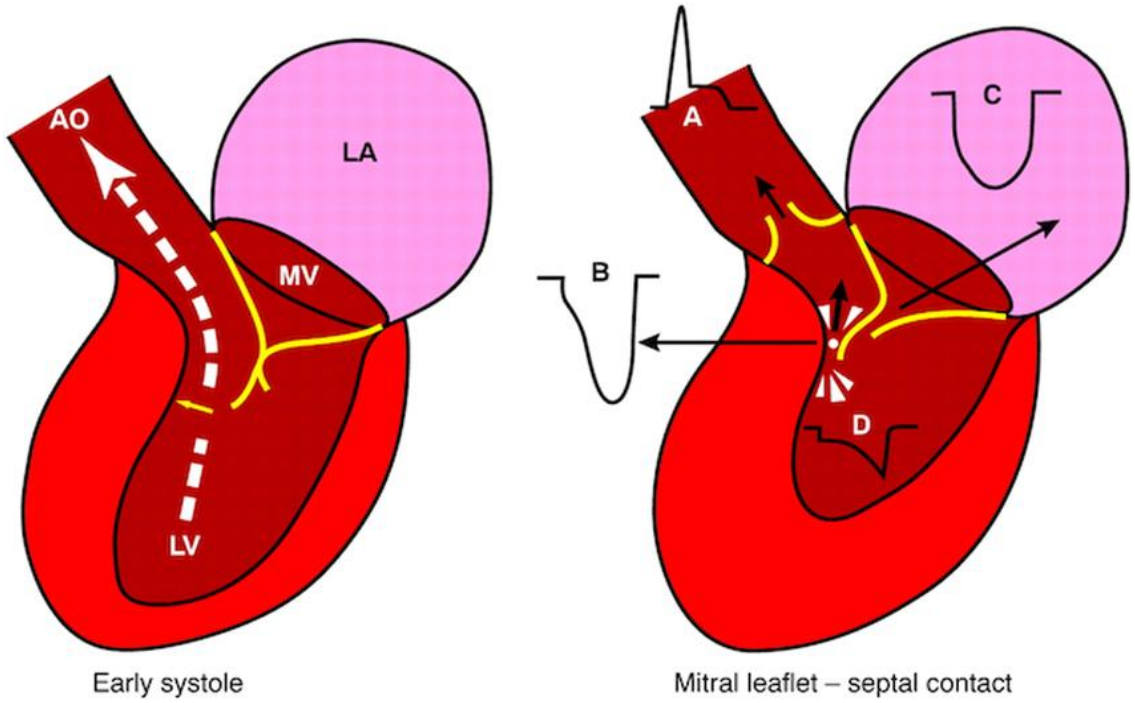
Yogasundaram H, Nikhanj A, Chatur S, Qi A, Hagen L, Bailey L, Khan A, Hopkin RJ, Fine NM, Jefferies JL, Oudit GY. Burden of Valvular Heart Disease in Patients with Fabry Disease. J Am Soc Echocardiogr. 2022 Feb;35(2):236-238. doi: 10.1016/j.echo.2021.09.013. Epub 2021 Oct 7. PMID: 34627970.

SAM-SYSTOLIC ANTERIOR MOTION

- **Mechanismus**
  - **„drag forces“** krevního toku posouvají mitrální cípy **anteriorně do LVOT**
  - Venturiho efekt **není hlavní mechanismus**
- **Anatomické predispozice**
  - **dlouhé cípy mitrální chlopně**
  - **anteriorní/apikální dislokace papilárních svalů**
  - **hypertrofie IVS**
  - úzké LVOT

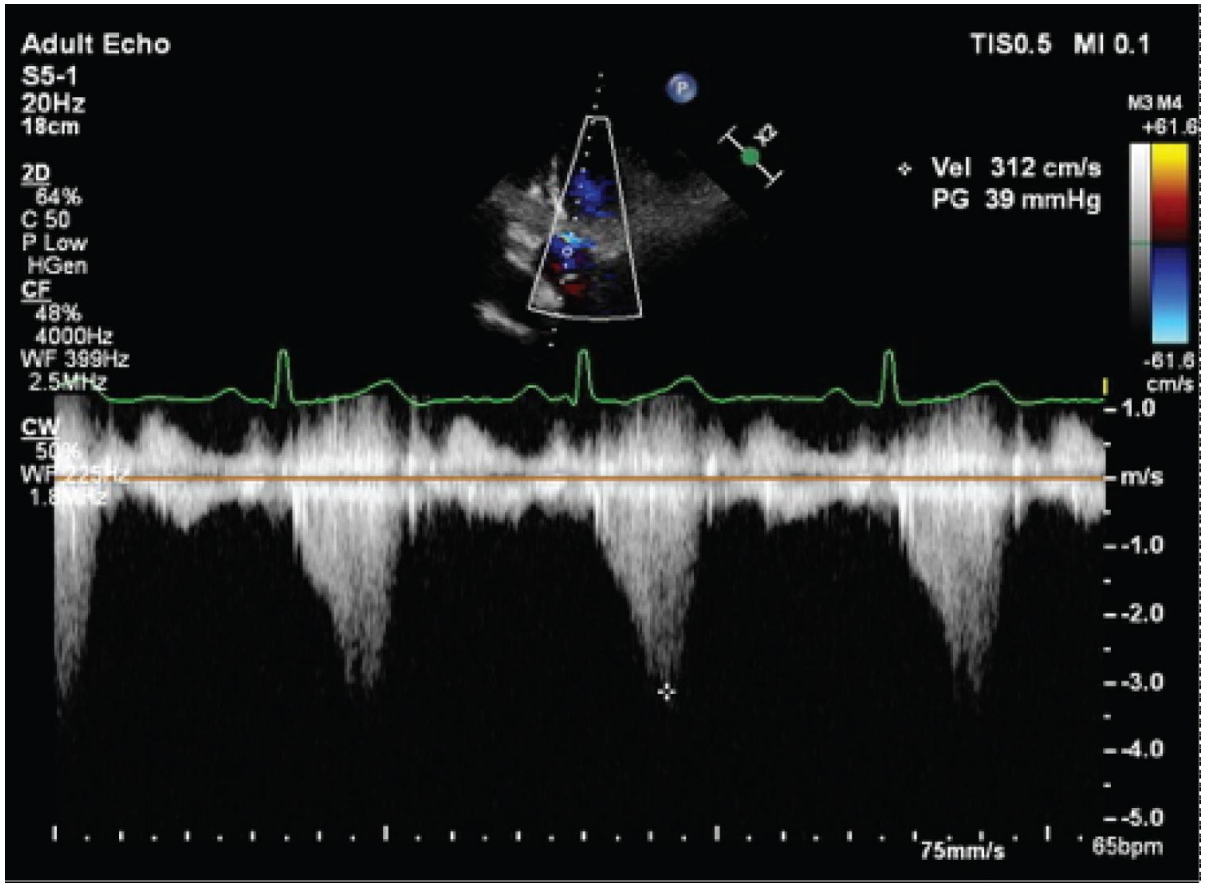


LVOTO Fabryho nemoci - Centrum FD VFN



Early systole

Mitral leaflet – septal contact



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Nejčastější chlopenní vada u FABRYHO CHOROBY je MITRÁLNÍ REGURGITACE

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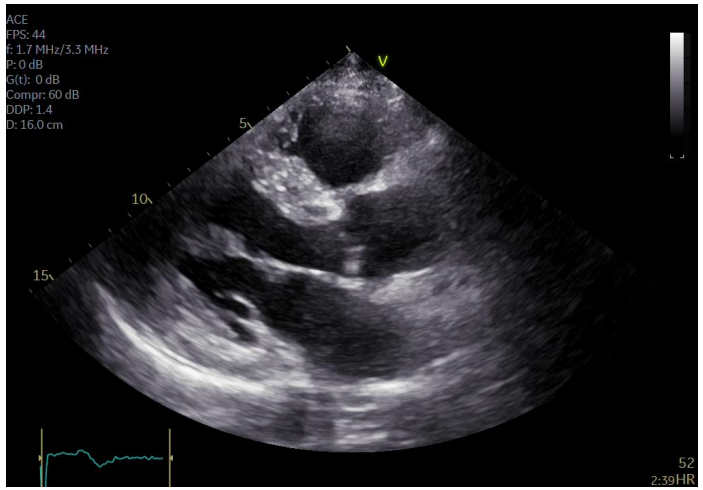

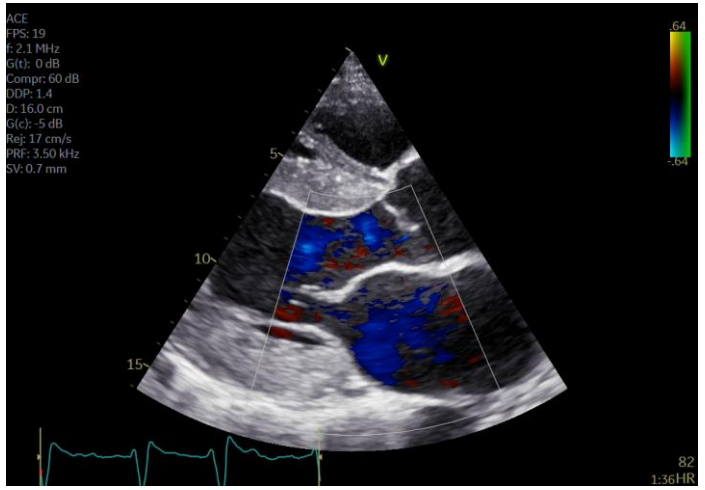

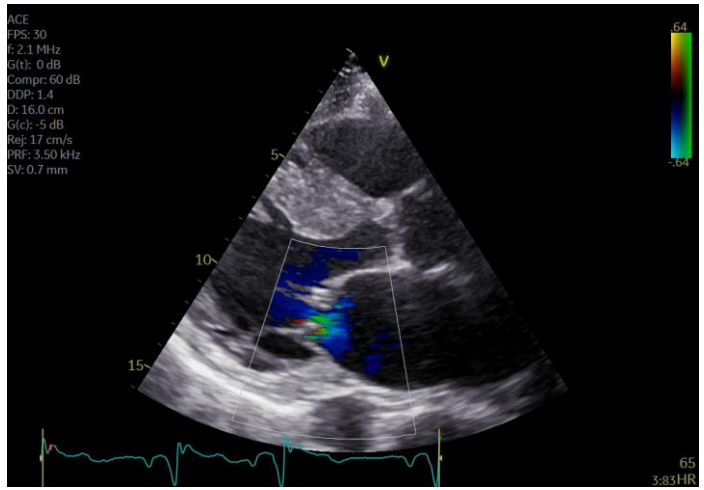

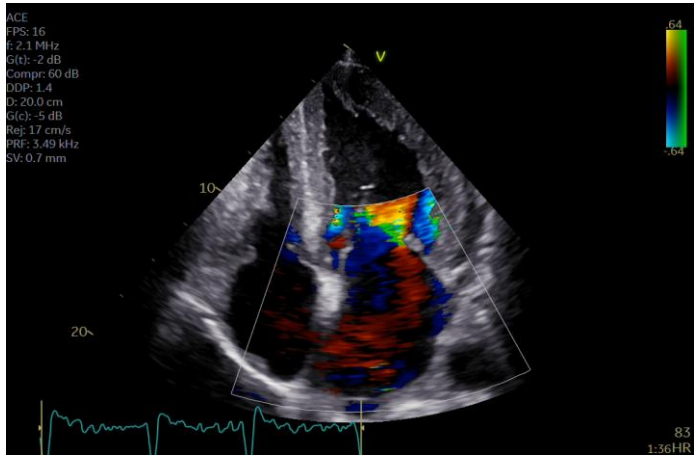

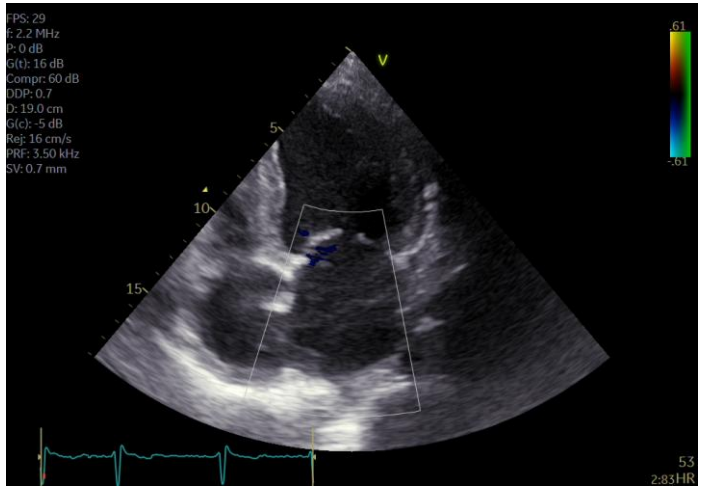

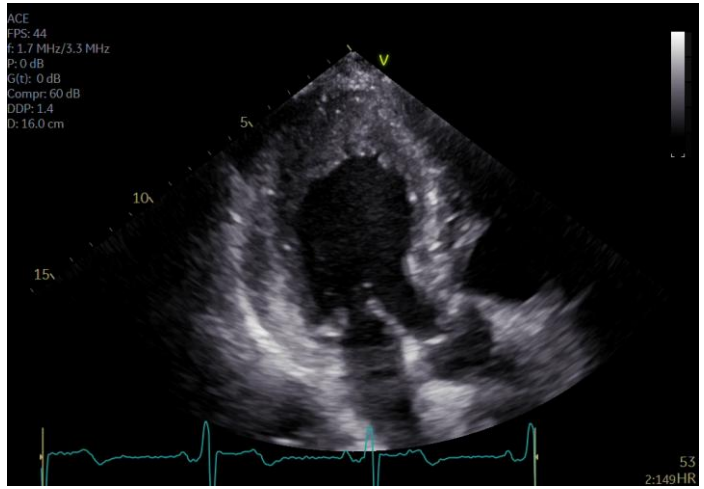

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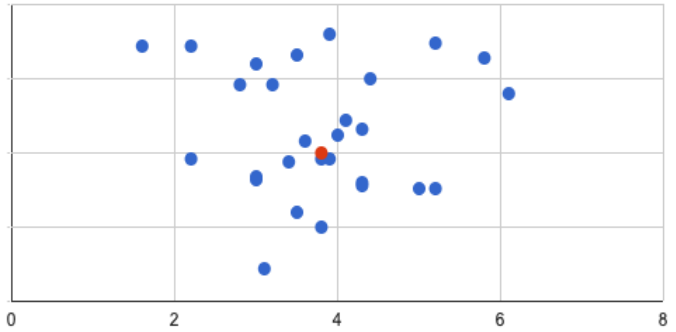
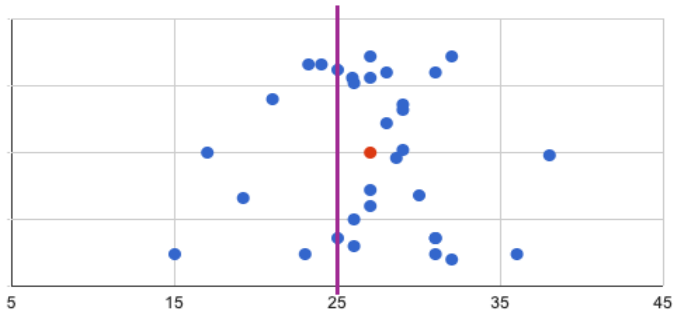
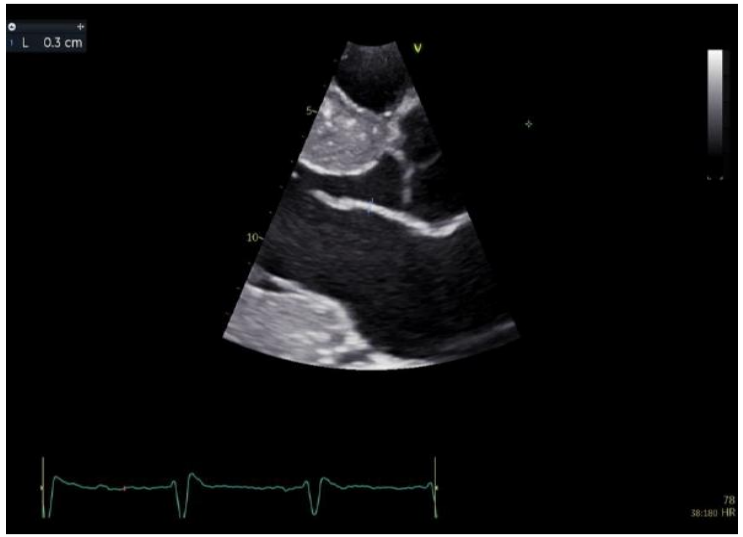
CENTRUM FABRYHO CHOROBY VFN

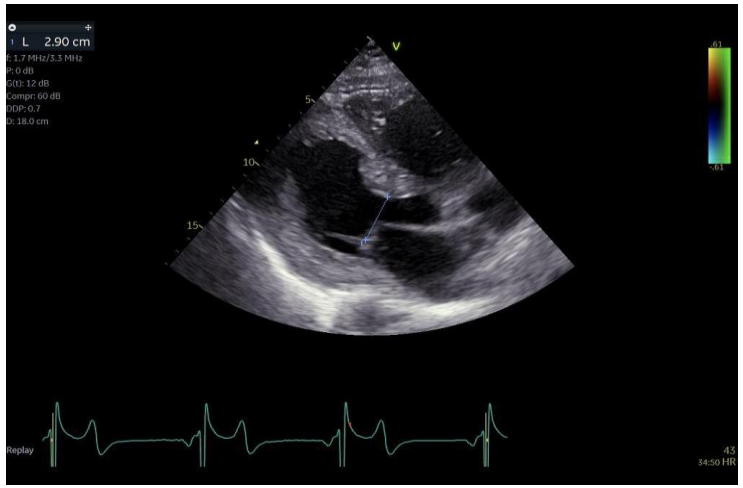
MVLT (mitral valve leaflets thickness)



C-SEPT (mm)







55 - MMR nebo SMR

120 - patogenní varianta

289 – celkem FD registr

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VŠEOBECNÁ FAKULTNÍ NEMOCNICE V PRAZE | 1. LÉKAŘSKÁ FAKULTA Univerzita Karlova

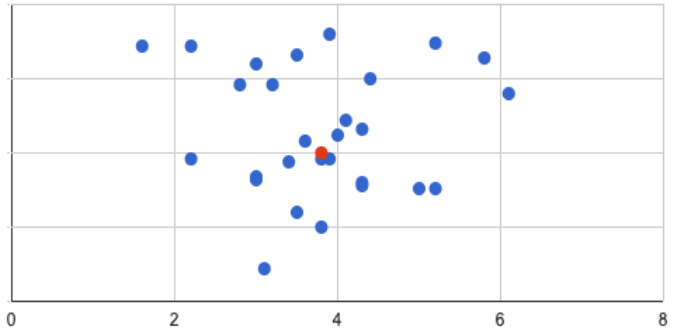
55 - MMR nebo SMR

CENTRUM FABRYHO CHOROBY VFN

120 - patogenní varianta

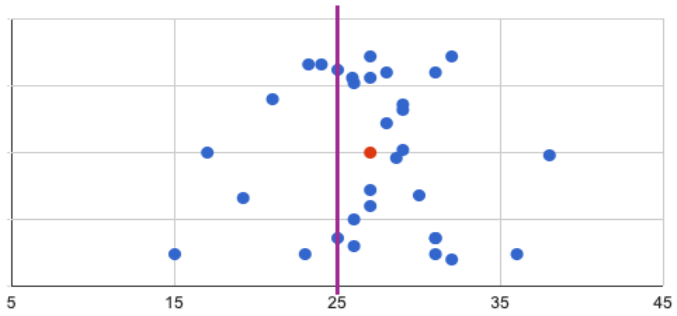
289 – celkem FD registr

MVLT (mitral valve leaflets thickness)



Unique	Min	Max	Mean	StDev	Sum	Percentile						
						0.05	0.10	0.25	0.50 Median	0.75	0.90	0.95
29	1.50	6.10	3.47	1.09	159.50	1.83	2.05	2.65	3.50	4.10	4.85	5.20

C-SEPT (mm)

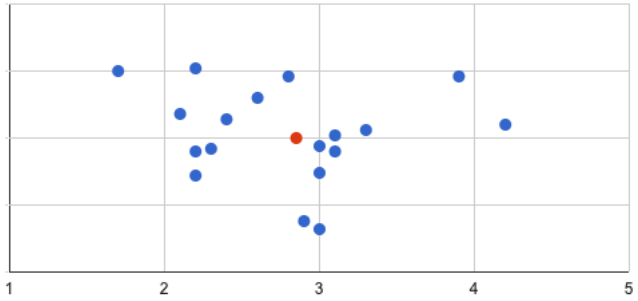


Unique	Min	Max	Mean	StDev	Sum	Percentile						
						0.05	0.10	0.25	0.50 Median	0.75	0.90	0.95
24	2.60	38	26.61	6.12	1330.4	16.95	19.08	24	27	31	32	34.65

AMBULANCE S HCM VFN

21 - MMR nebo SMR

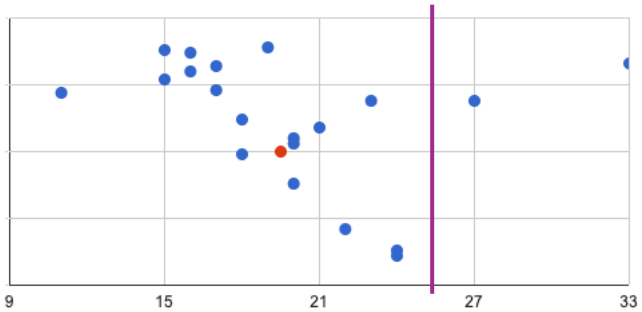
MVLT (mitral valve leaflets thickness)



mm

Unique	Min	Max	Mean	StDev	Sum	Percentile						
						0.05	0.10	0.25	0.50 Median	0.75	0.90	0.95
13	1.70	4.20	2.78	0.640	50	2.04	2.17	2.23	2.85	3.08	3.48	3.95

C-SEPT (mm)



mm

Unique	Min	Max	Mean	StDev	Sum	Percentile						
						0.05	0.10	0.25	0.50 Median	0.75	0.90	0.95
13	11	33	19.80	4.88	396	14.80	15	16.75	19.50	22.25	24.30	27.30

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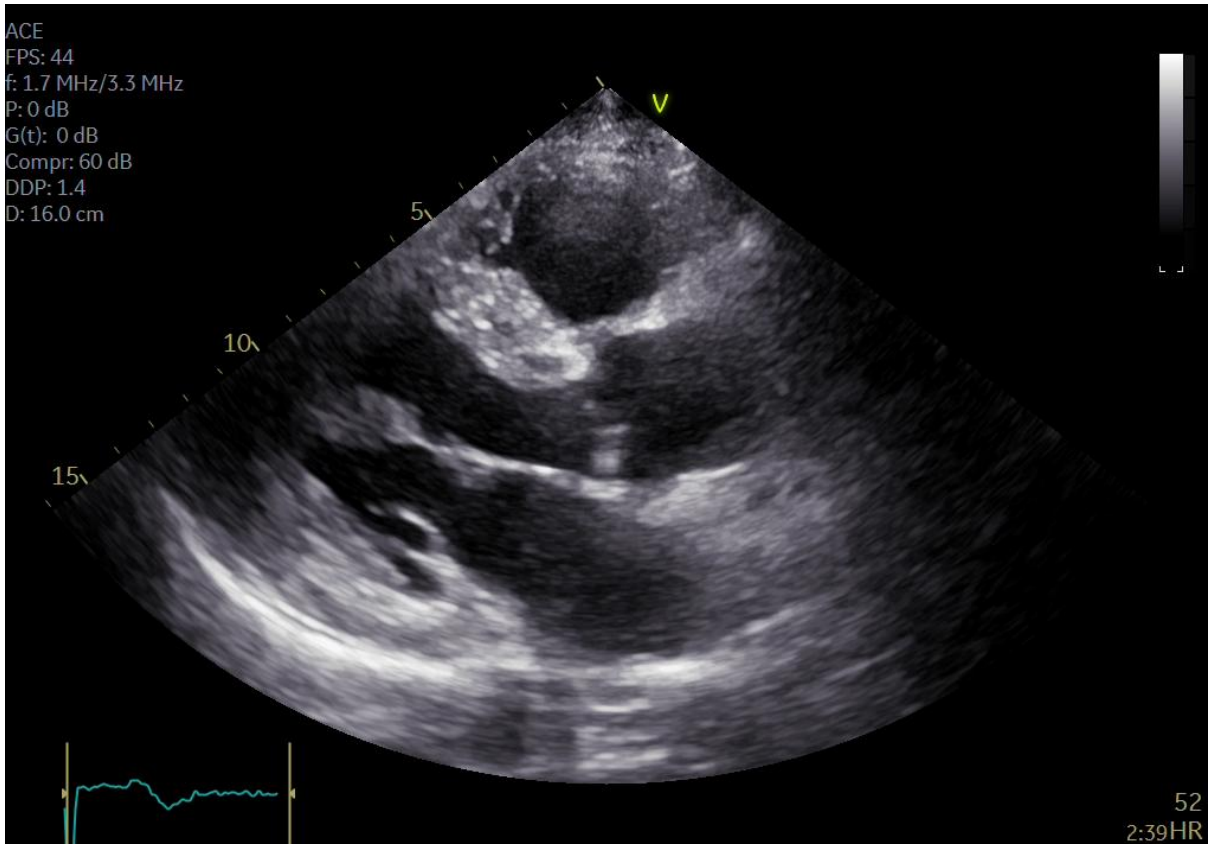
SROVNÁNÍ SAM u FD a HCM

FABRYHO CHOROBA



HCM

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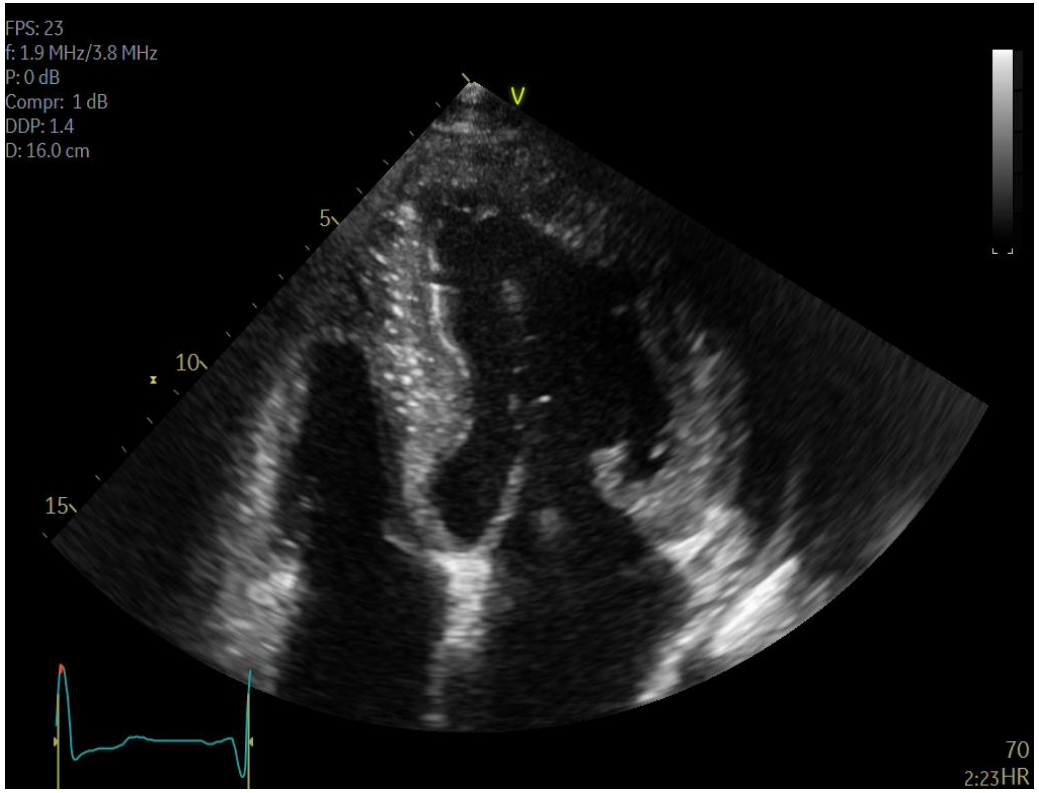
LVOTO - left ventricular outflow tract obstruction (FD)



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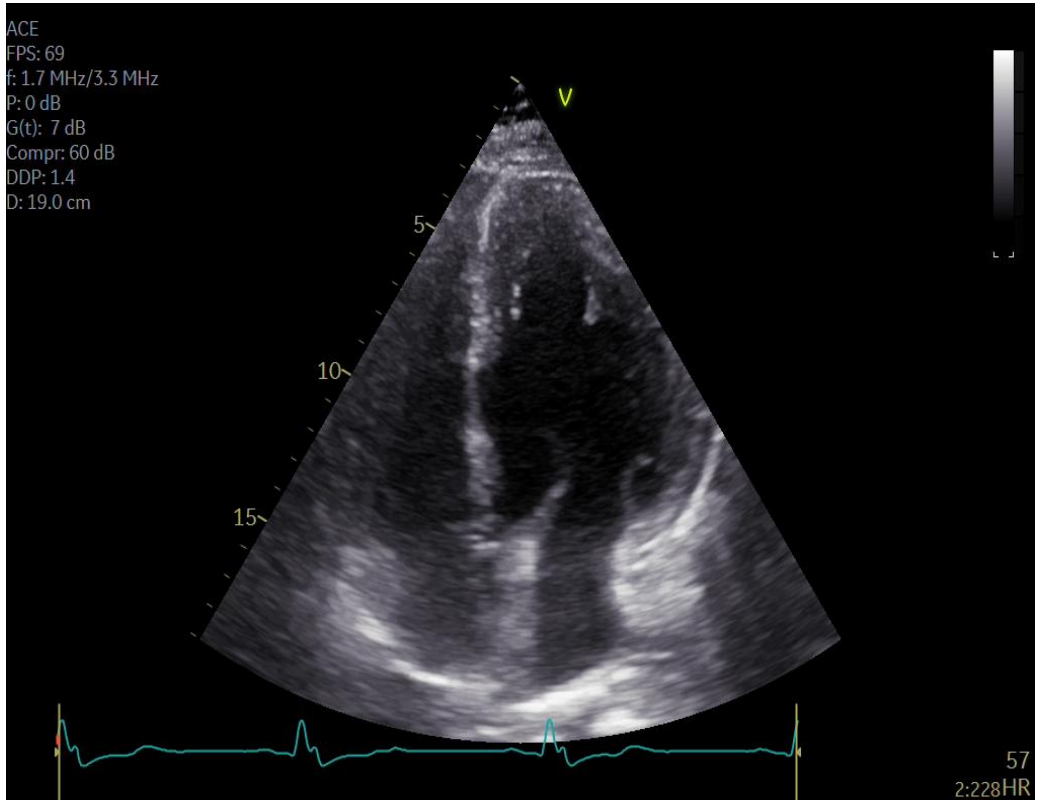
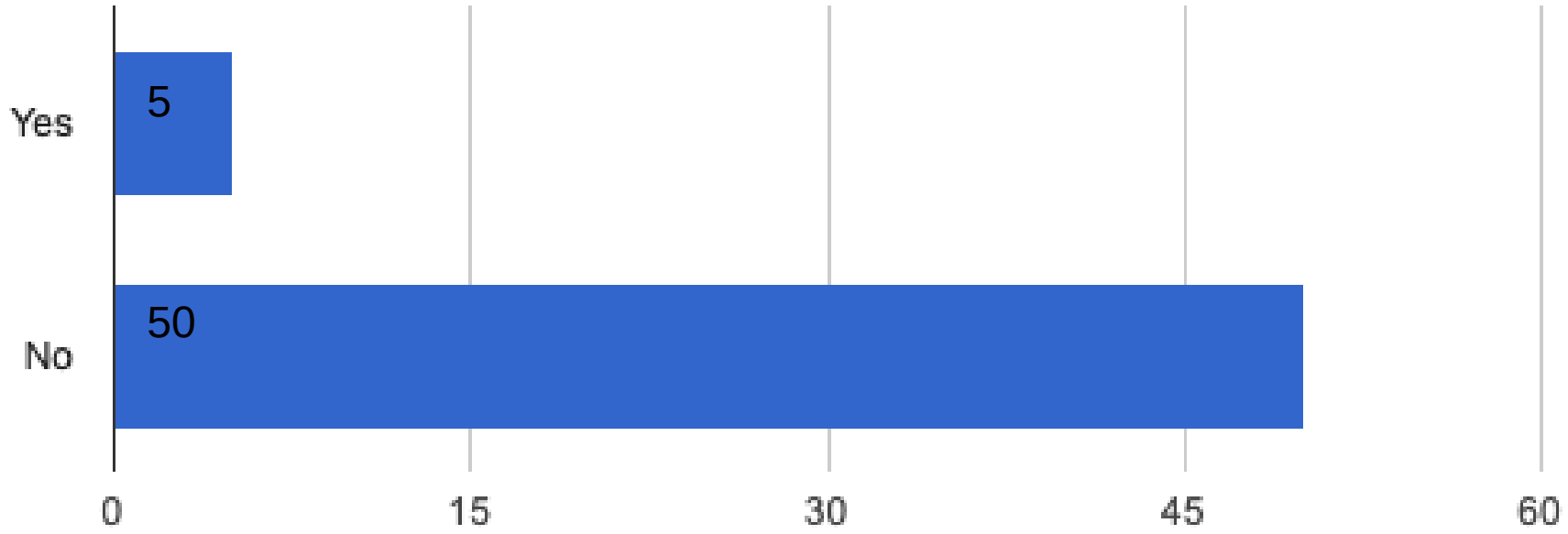


PTSMA

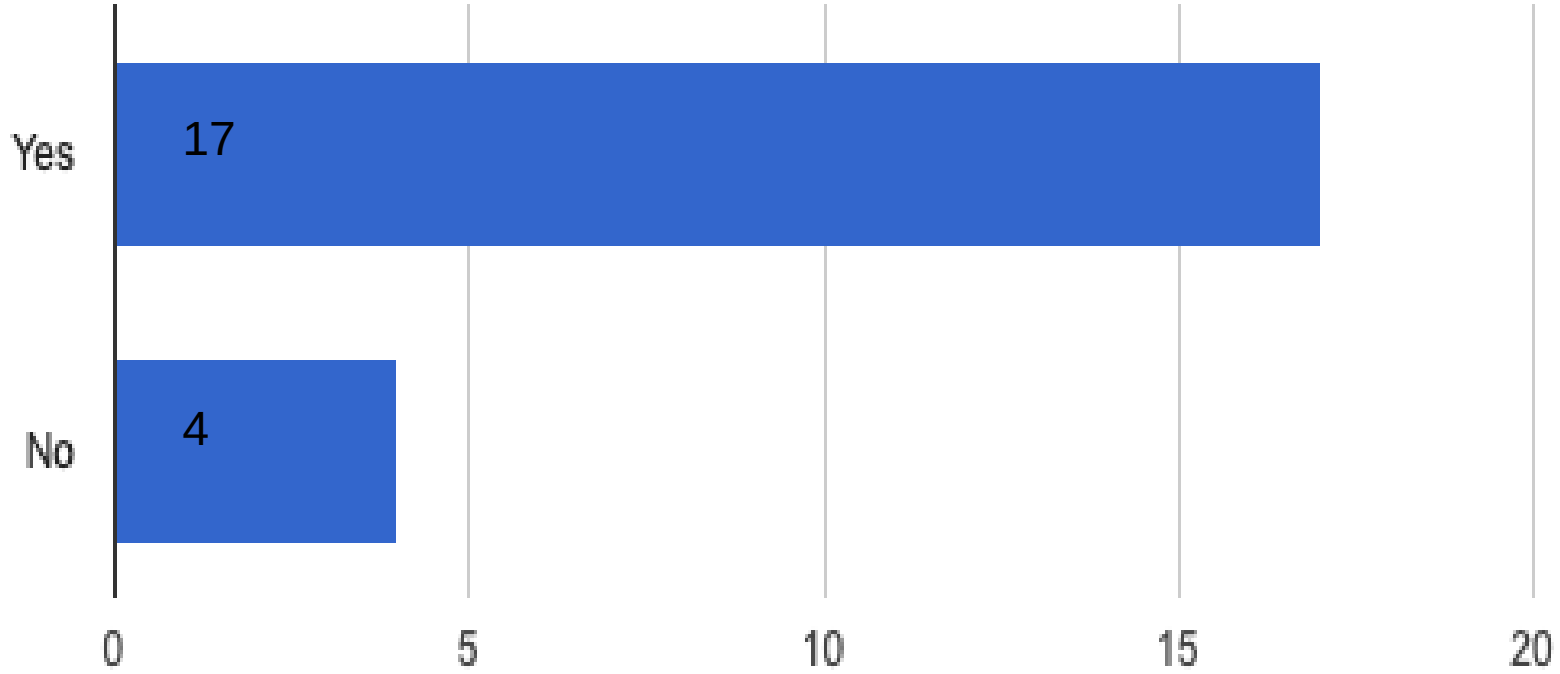
CELKOVÝ POČET pacientů s Fabryho chorobou a středně významnou nebo významnou MR: 55

LVOTO v klidu Yes (5, 9.1%), No (50, 90.9%)



CELKOVÝ POČET pacientů s HCM a středně významnou nebo významnou MR: 21

OBSTRUKCE LVOT v klidu: Yes (17, 81.0%), No (4, 19.0%)



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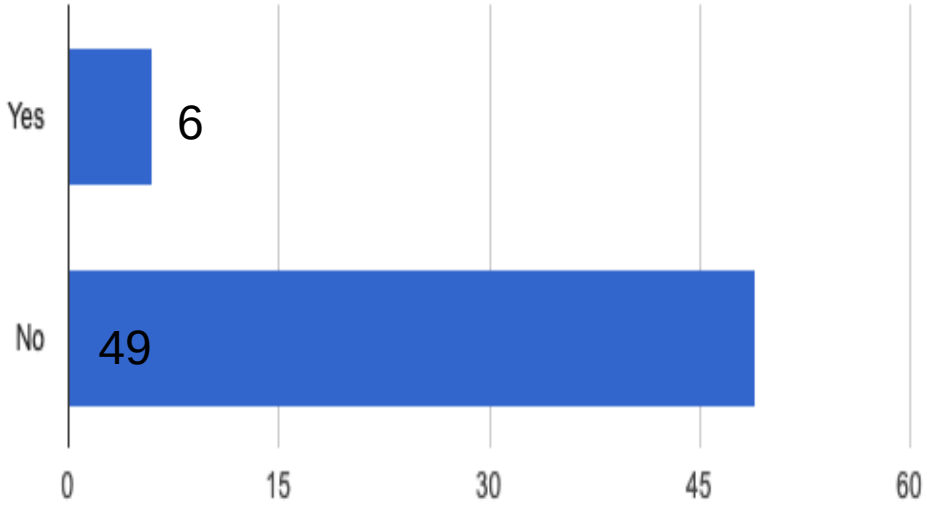
SAM u pacientů se středně významnou a významnou mitrální regurgitací

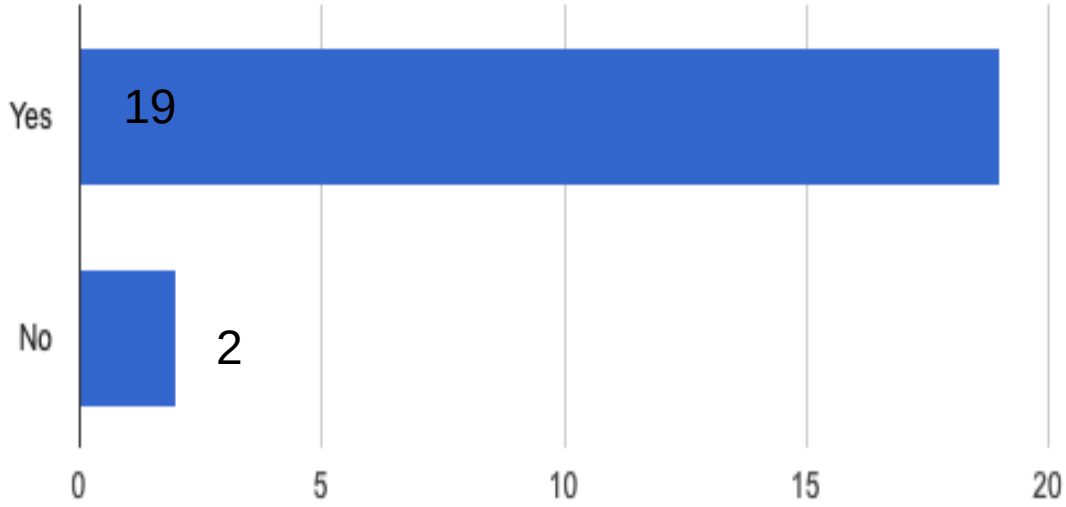
FABRYHO CHOROBA

Yes (6, 10.9%), No (49, 89.1%)



SARKOMERICKÁ HCM

Yes (19, 90.5%), No (2, 9.5%)



ZÁVĚR:

- SAM je výrazně častější u pacientů se sarkomerickou hypertrofickou kardiomyopatií než u pacientů s Fabryho chorobou
- Patofyziologická podstata tohoto jevu má více příčin
- Z dosud získaných dat se domníváme, že restrikce u střádavé choroby (FD) je paradoxně ochranou vzniku SAM

VŠEOBECNÁ FAKULTNÍ NEMOCNICE V PRAZE

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DĚKUJI ZA POZORNOST…

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