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Puzzling cases in cardiac function: Read with the experts

All that Glitters is not Gold

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18-Year-Old Male

- Patent ductus arteriosus dx at birth
- Echo at 5 months showed closure of PDA but severe LVH
- Albuterol treatment caused tachycardia

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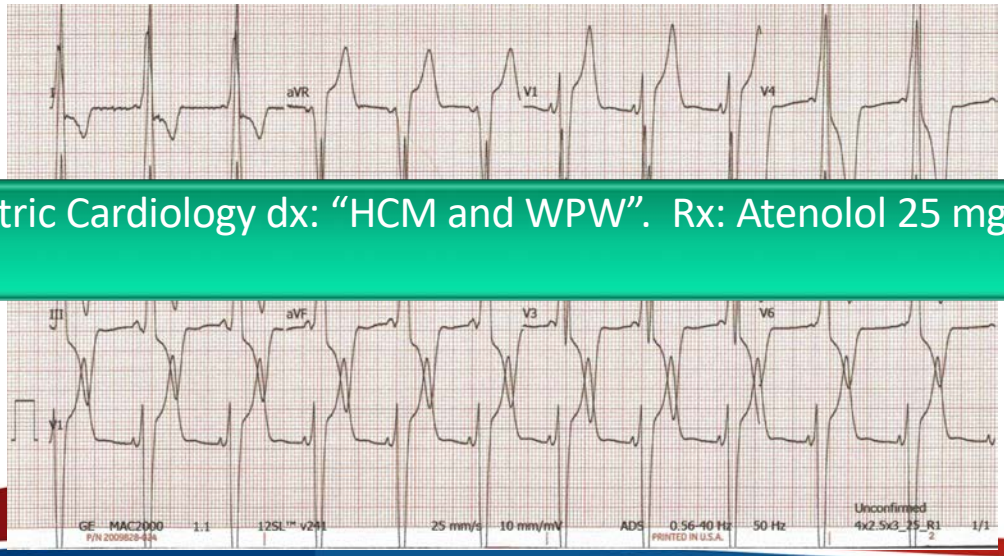
ECG



Pediatric Cardiology dx: "HCM and WPW". Rx: Atenolol 25 mg daily

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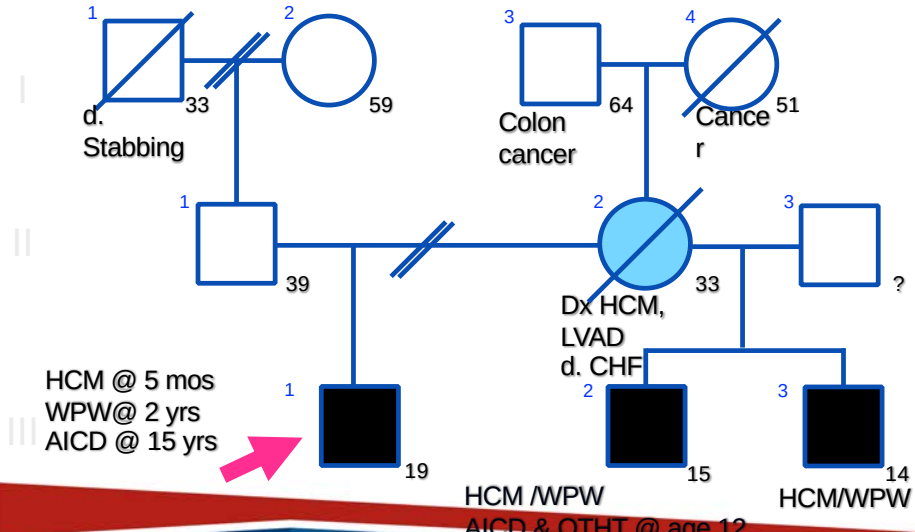
Family Pedigree



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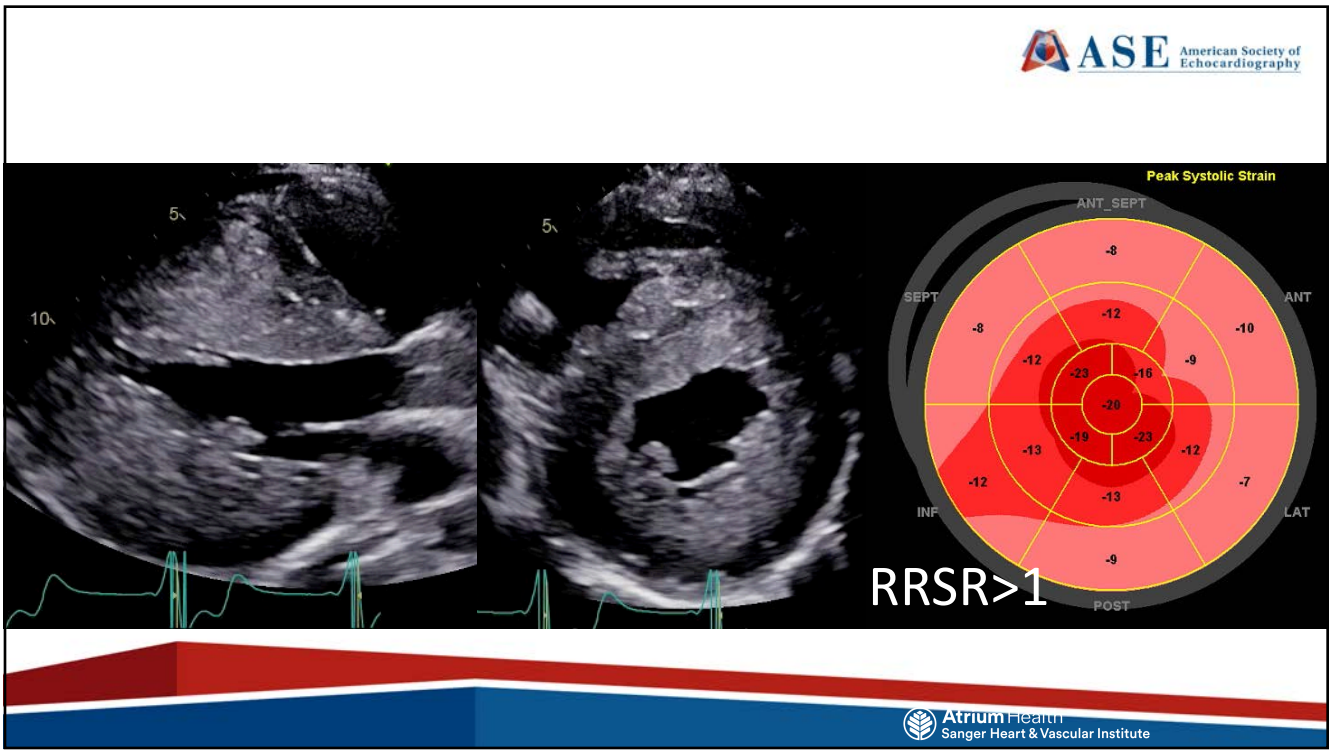

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What is the diagnosis?

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1. Agree with the pediatric cardiologist: HCM
2. Apical sparing pattern on strain: this is amyloid
3. Something else

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Cardiomyopathies Associated With W-P-W Syndrome and Short PR

Syndrome	Type of CMP
Danon disease	**HCM**
PRKAG2 syndrome	**HCM**
Fabry	**HCM**
Pompe disease	HCM
Duchenne/Becker MD	DCM
MELAS syndrome	HCM, DCM
Kearns-Sayre syndrome	DCM
Leigh syndrome	HCM, DCM
MERRF syndrome	HCM, DCM

Porto AG et al. *Circ Arrhythm Electrophysiol.* 2016;9:1-8.

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RESULTS AND INTERPRETATION

Test Name
Hypertrophic Cardiomyopathy Gene Panel *(MYBPC3, MYH7, TNNI3, TNNT2, TPM1, ACTC1, MYL2, MYL3, LAMP2, GLA, PRKAG2, TTR)*

Result
Pathogenic variant in *LAMP2* gene

Gene	Exon	Variant	Zygosity	Classification	Inheritance	Parental origin
LAMP2 NM 002294.2	6	c.763_768delinsTGAAGT, (p.Asn255*)	Homozygous	Pathogenic	X-linked dominant	Likely maternal

Lysosomal glycogen storage disease

Lysosomal-associated membrane protein 2 (LAMP2) transporter protein deficiency

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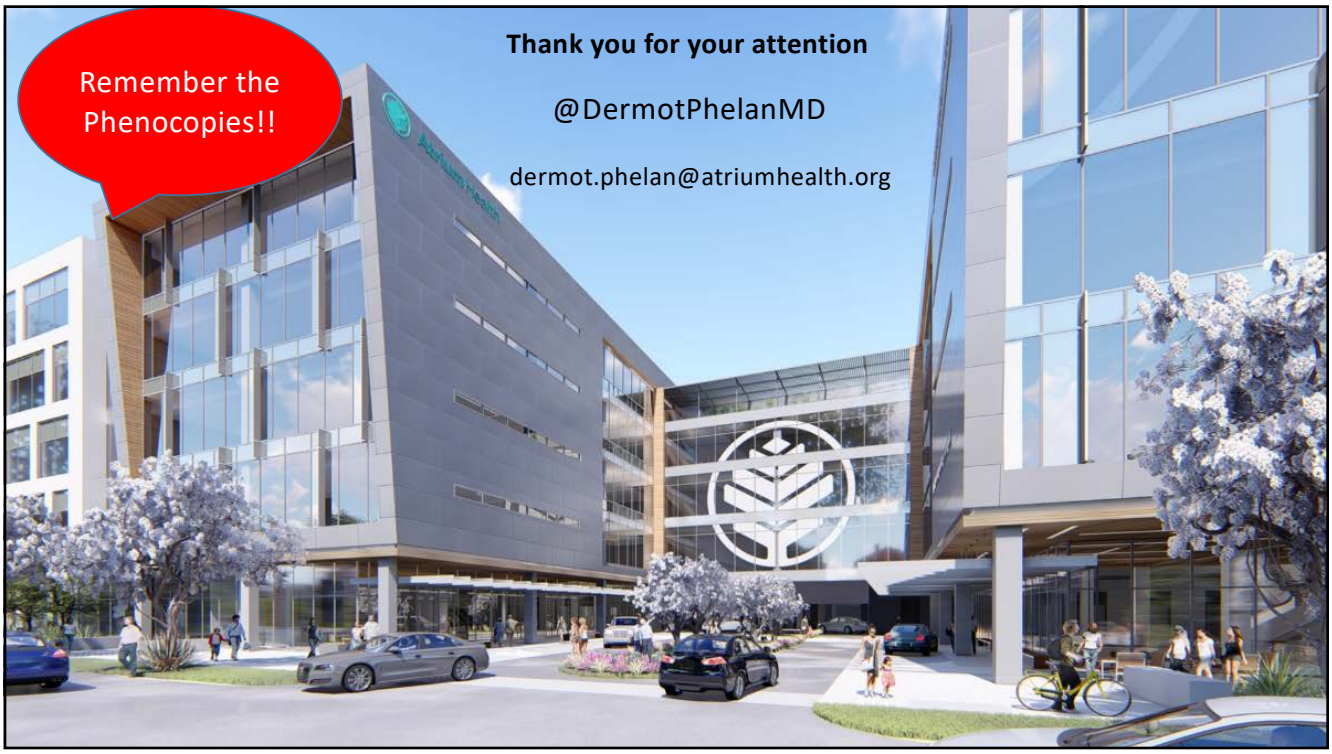
Remember the Phenocopies!!
Thank you for your attention
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