
Minneapolis Heart Institute Foundation
GRAND ROUNDS

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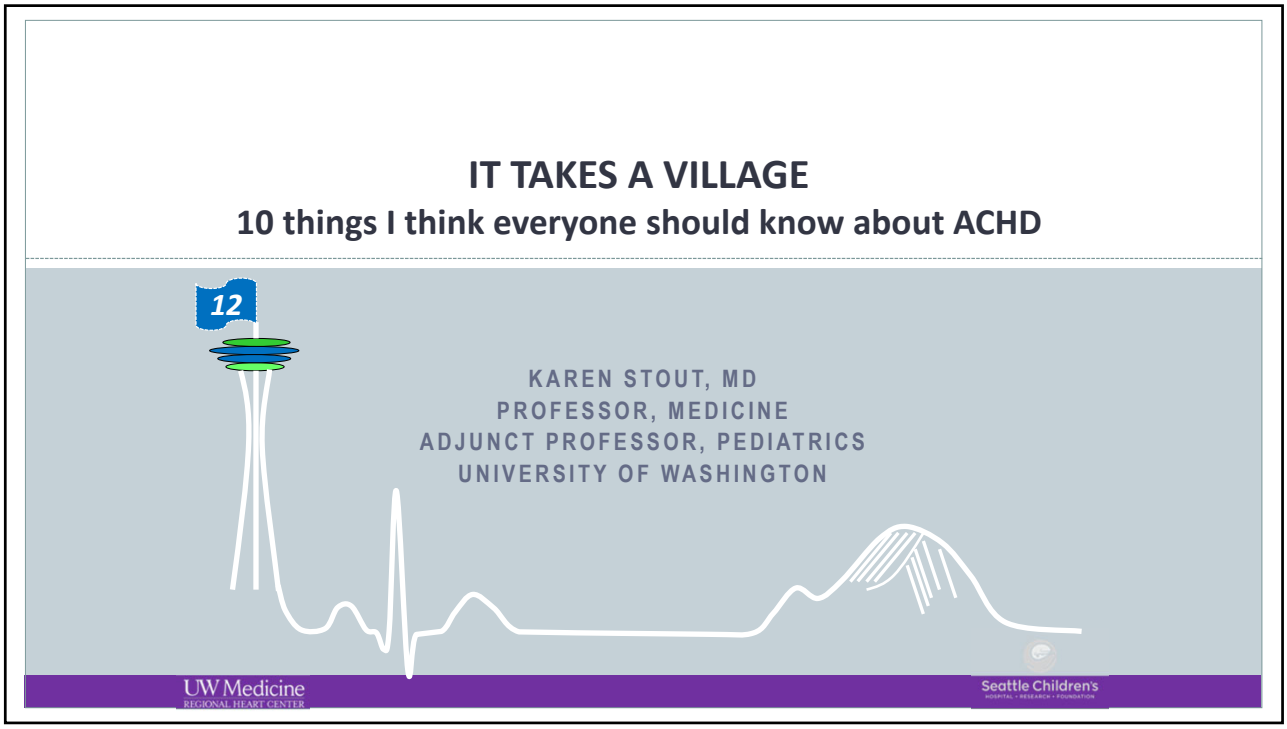
IT TAKES A VILLAGE
10 things I think everyone should know about ACHD
12
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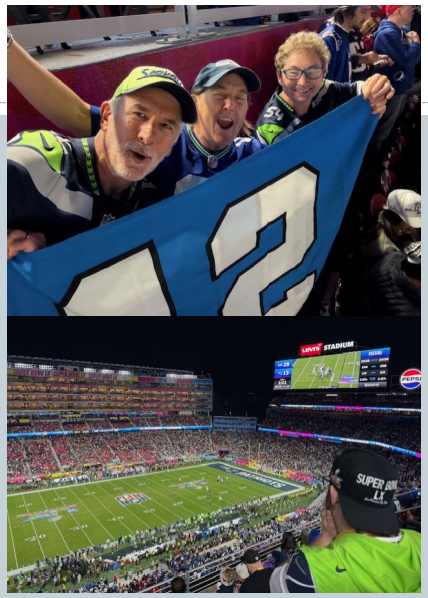
ADJUNCT PROFESSOR, PEDIATRICS
UNIVERSITY OF WASHINGTON
UW Medicine
REGIONAL HEART CENTER
Seattle Children's

2

I HAVE NO DISCLOSURES

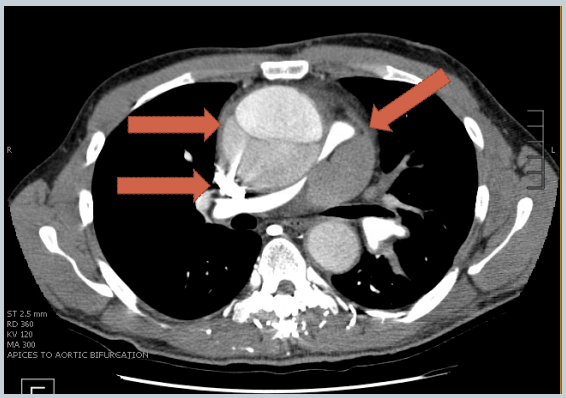


3

One Summer Sunday as a Cardiology Fellow

- 27 year old man with a history of coarctation of the aorta and bicuspid aortic valve
- Coarctation repair at age 4
- Mechanical AVR at age 16
- Followed by PCP for hypertension
- No cardiology visit since age 19

- Seen in ED for chest pain and 'blue head'



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Aspirations

- **Why know about ACHD?**
- **What makes ACHD unique?**
- **What might be more familiar?**

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#1 ACHD IS COMMON. YET NOT?

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CONGENITAL HEART DISEASE
not encompassing all 'inherited' heart disease

DEFINITION

Heart disease evident since birth

Think holes, abnormal connections, abnormal valves

Atrial Septal Defect	Tetralogy of Fallot
Ventricular Septal Defect	Transposition of the great arteries
Atrioventricular Septal Defect	Truncus arteriosus
Congenital valvular stenosis	Valve atresia
Congenital valvular regurgitation	Single ventricle
Coarctation of the aorta	Anomalous pulmonary venous return
Interrupted aortic arch	Anomalous coronary artery origin

Seattle Children's

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Congenital heart disease is the most common type of birth defect

1% of babies are born with CHD	40,000 births per year in the US	25% of those have a critical, life threatening defect

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10,000 babies born in the US with critical congenital heart disease annually

Before 1950s
- Most died in infancy
- Survivors had severe limitations

1950s-1980s
- Rapid improvement in surgical technique
- Improvement in medication/imaging
- Many survived to adulthood

"Modern" era
- Death during childhood is rare
- Expected survival to age 18 >95% for all CHD
- 70% of those with CHD turning age 18 will reach age 70

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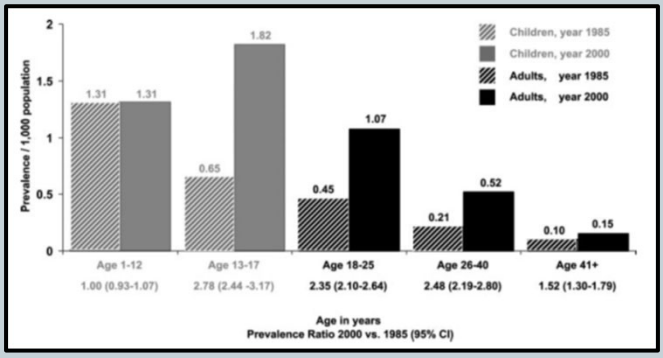
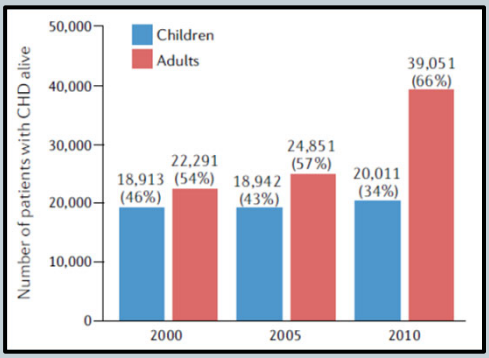
PREVALENCE OF CHD BY AGE



Marelli et al *Circulation* 2007



Liu et al *Nature Reviews* 2023

>1,500,000 Adults with congenital heart disease in the US

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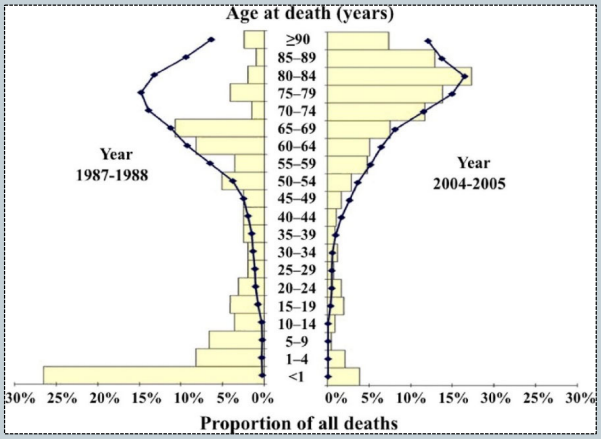
Congenital heart disease is no longer a problem of children

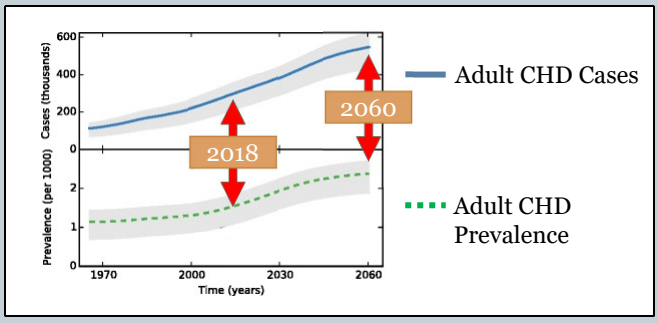
Expected life span more closely mirrors general population and fewer die during childhood



Khairy *J Am Coll Cardiol*.2010;56:1149–57

11

The population will continue to grow



Benziger, Stout *Popul Health Metr*. 2015

>1.5 million ACHD patients in the US

Compare with…

468,000 on dialysis (2016)
400,000 with cirrhosis (2015)
120,000 with sickle cell disease (2010)
30,000 with Cystic Fibrosis (2017)

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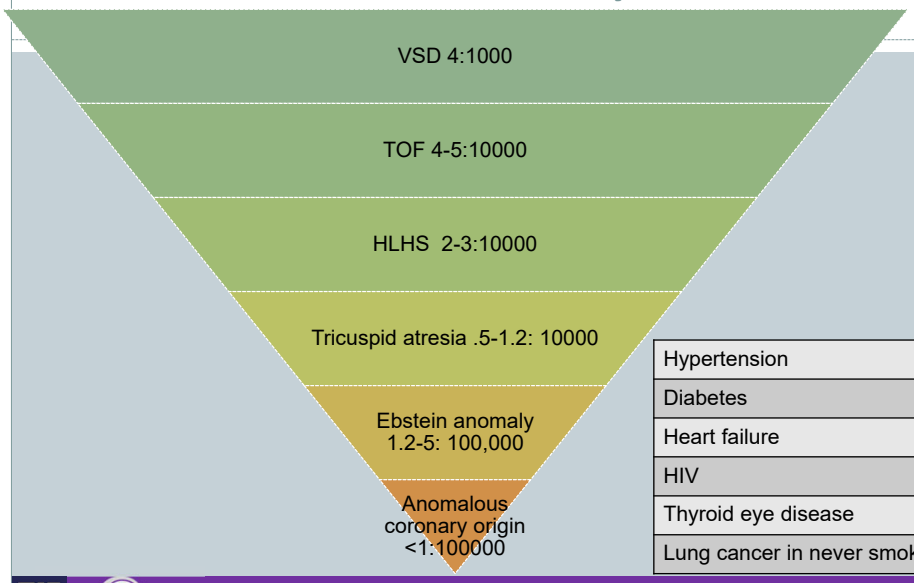
If it's so common, why does it feel so rare???



Hypertension	45% of adults
Diabetes	10% of adults
Heart failure	8-70/1000, 6 million in US
HIV	1.2 million in US
Thyroid eye disease	9/10,000
Lung cancer in never smoker	11-13/100000 person years

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#2 ACHD IS 'LEGIT' AS A SPECIALTY, YET NOT ENOUGH ACHD DOCS

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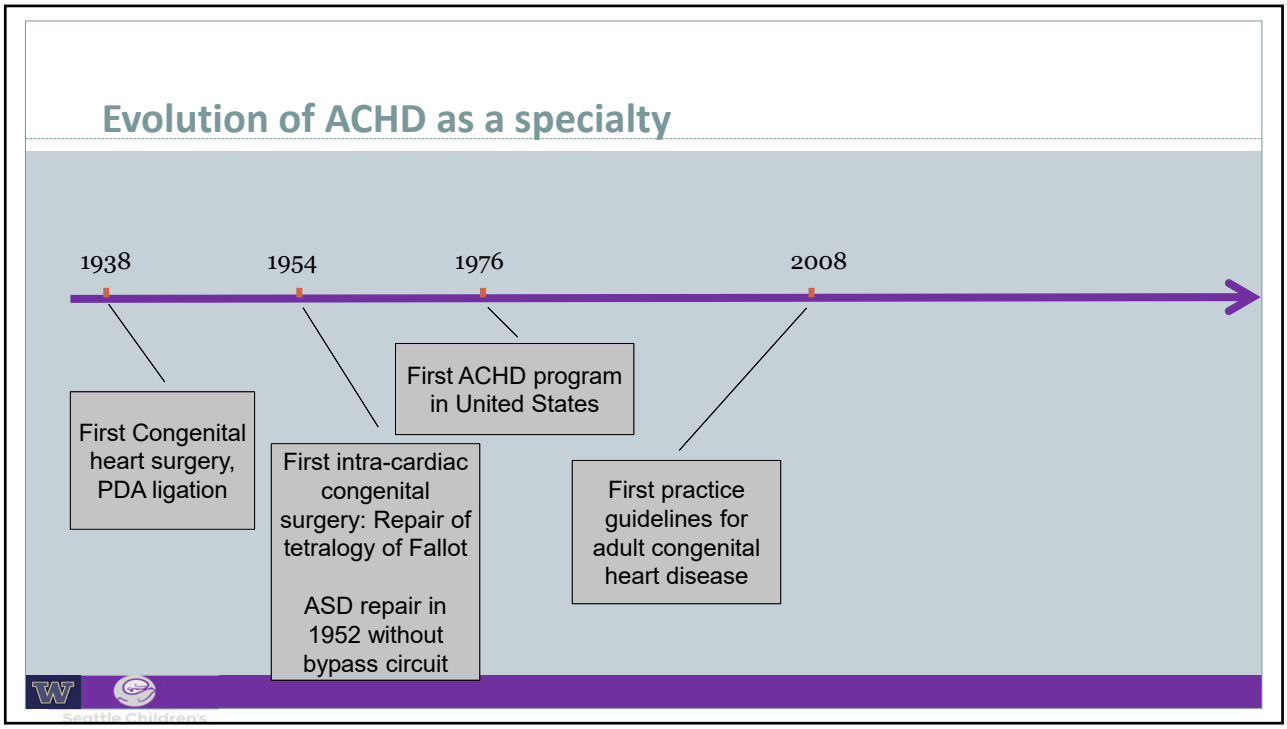
Evolution of ACHD as a specialty
1938
1954
1976
2008
First Congenital heart surgery, PDA ligation
First intra-cardiac congenital surgery: Repair of tetralogy of Fallot
ASD repair in 1952 without bypass circuit
First ACHD program in United States
First practice guidelines for adult congenital heart disease

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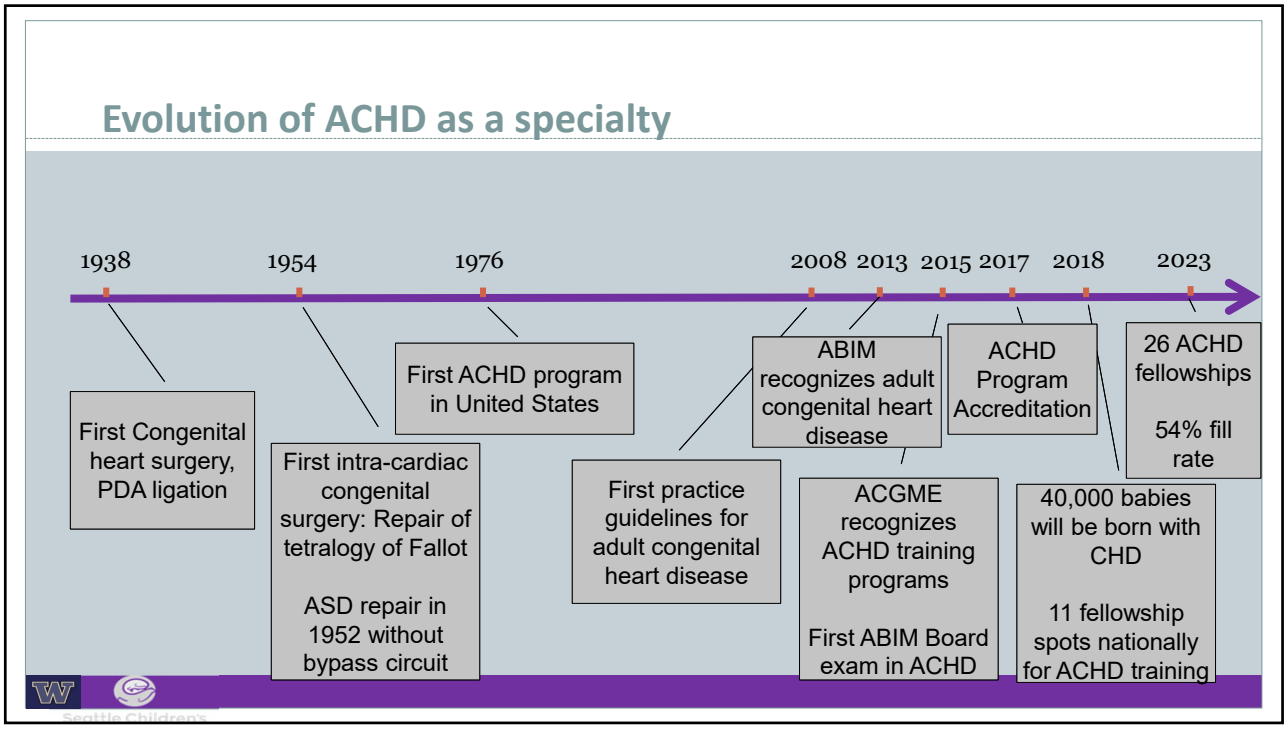
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First ACHD program in United States
First practice guidelines for adult congenital heart disease
ABIM recognizes adult congenital heart disease
ACGME recognizes ACHD training programs
First ABIM Board exam in ACHD
ACHD Program Accreditation
40,000 babies will be born with CHD
11 fellowship spots nationally for ACHD training
26 ACHD fellowships
54% fill rate

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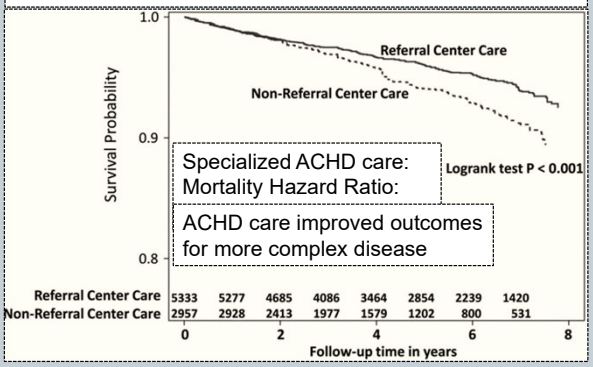
ACHD centers improve care for those with more complex CHD

But not enough ACHD docs to do the job

95% of general cardiologists see ACHD patients	<10% of ACHD patients are seen by ACHD doctors	80% of cardiologists reference the guidelines for care advice

Fernandes SM, *J Am Coll Cardiol*, 60(23) 2012

71,500 adult congenital patients



Marelli A, *Circulation*, 129(18) 2014

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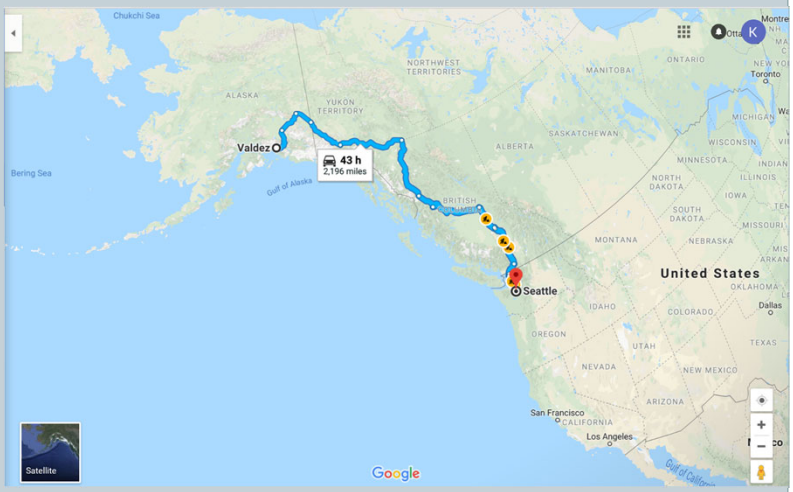
Michael

- 32 year old man with tetralogy of Fallot
- In for routine ACHD follow up evaluation
- Occasional palpitations
- Doesn't feel as in shape as in years past
- **Lives FAR from the nearest ACHD program**



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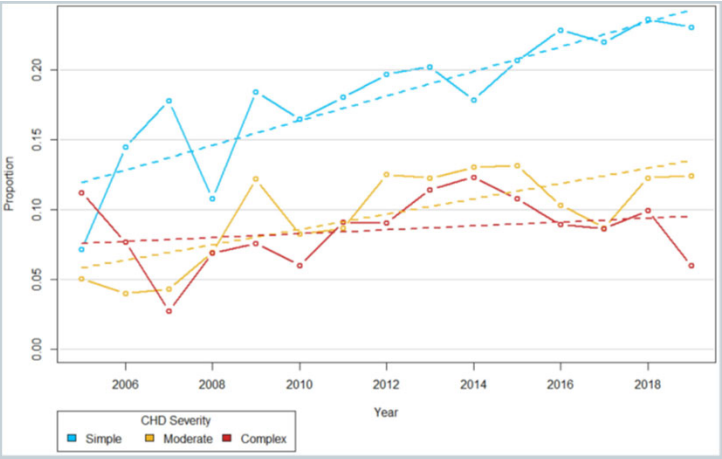
Gaps in Care are on the rise



Zaidi et al *J Amer Heart Assoc* 2024

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What do the guidelines say about who should provide care for ACHD patients?

Recommendations for Delivery of Care
Referenced studies that support recommendations are summarized in Online Data Supplements 3, 4, and 5.

COR	LOE	Recommendations
I	B-NR	1. Patients with ACHD AP classification IB-D, IIA-D, and IIIA-D* should be managed in collaboration with an ACHD cardiologist (S3.3-1).
I	C-LD	2. Cardiac surgery, catheter-based interventional cardiac procedures, and electrophysiological procedures involving congenital heart lesions in patients with ACHD should be performed by operators with expertise in CHD procedures and in collaboration with an ACHD cardiologist (S3.3-1, S3.3-2).

*See Tables 3 and 4 for details on the ACHD Anatomic and Physiological classification system.

Stout et al J Amer Coll Card 2018

Recommendations for the ACHD Program and Cardiologists
Referenced studies that support recommendations are summarized in the Evidence Table.

COR	LOE	Recommendations
1	B-NR	1. Patients in ACHD AP classes IB–D, IIA–D, and IIIA–D should be evaluated and managed by or in collaboration with an ACHD cardiologist and, when indicated and accessible, an ACHD program to improve outcomes.[1–3]
1	B-NR	2. Patients in ACHD AP class IA should have at least 1 evaluation by an ACHD cardiologist to develop a plan of care.[1–3]

Gurvitz, Krieger et al J Amer Heart Assoc and J Amer Coll Card 2025

20

The population is continuing to outpace the available expertise

DON'T CONFUSE HOPE FOR A PLAN

We Need

More providers aware of ACHD

More ACHD Cardiologists

More resources to help provide care

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#3: WHAT ARE OUTCOMES IN ACHD?

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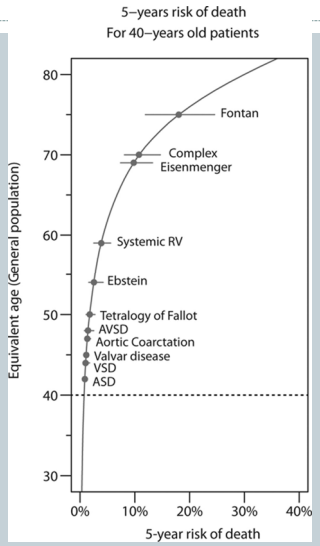
Most patients with ACHD do well – but if not, what happens? what are we trying to prevent?

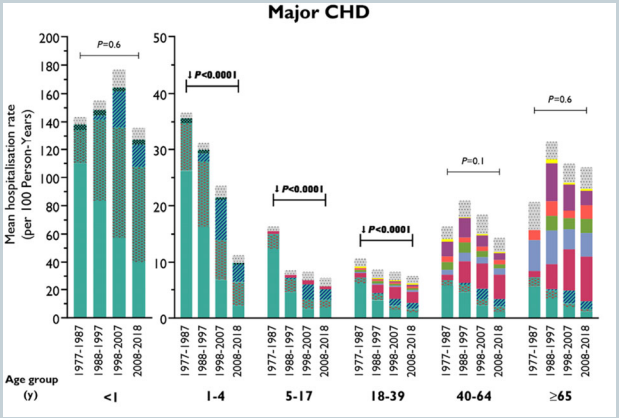
- Arrhythmias
- Heart failure
- Exercise intolerance
- Endocarditis
- Pulmonary hypertension
- Liver disease
- Renal disease
- Erythrocytosis

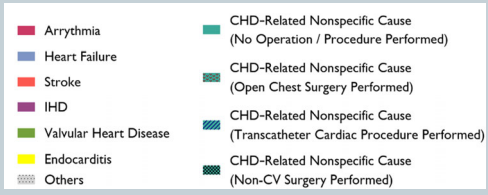


Diller GP, et al Circulation. 2015

23

Causes of hospitalizations by age and era Difference in kids vs. adults

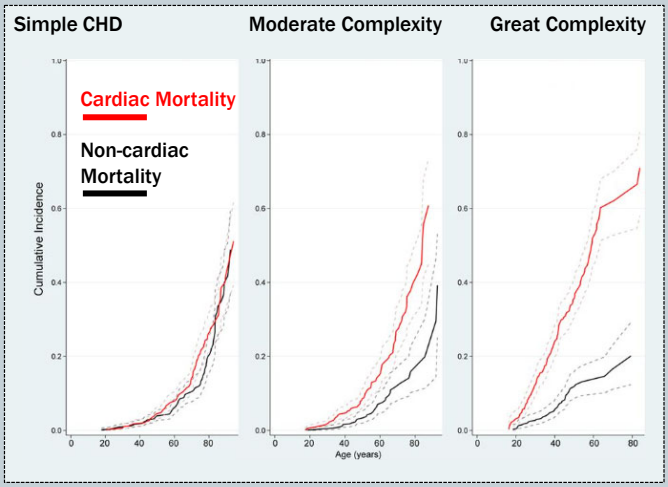


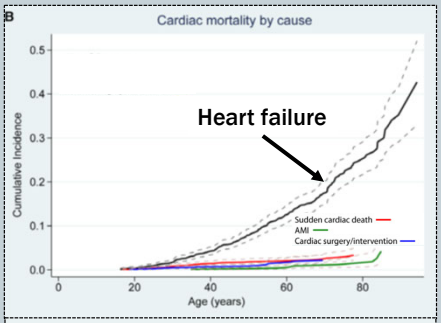


Lim, et al J Am Heart Assoc 2024

24

Cardiac causes drive mortality in most CHD patients





Heart failure remains the dominant cause of ACHD death in the modern era

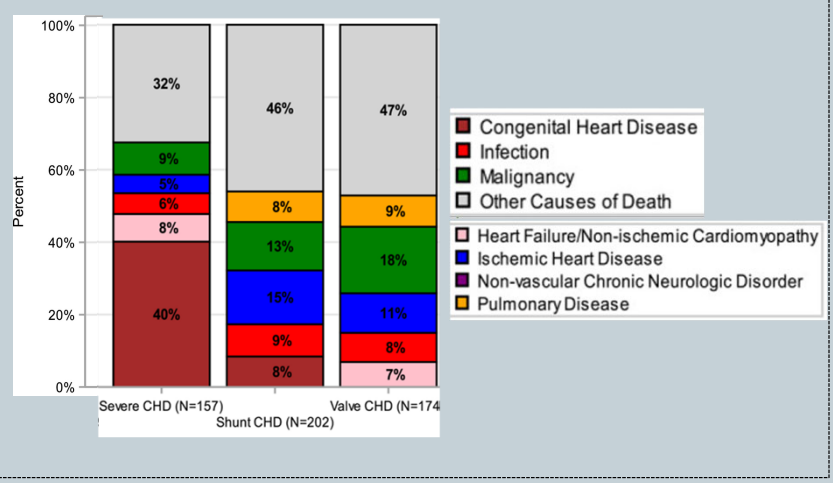
Diller GP, et al Circulation. 2015

25

Complex ACHD: CHD mortality dominates
Simpler ACHD: ASCVD, infection, cancer

628 patients who died with CHD

- Median age of death 64 years
  - Severe CHD : 46yrs
  - Shunts 65 yrs
  - Valvular CHD: 73 yrs



Goldstein et al. J Am Heart Assoc. 2020

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#4 WHAT ABOUT "AGE" RELATED DISEASES?

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Atherogenic Risk Factors Vary in ACHD Compared to General Population

Hypertension

- More prevalent across all diagnoses
- Most common coarctation, Williams syndrome

Dyslipidemia

- Lower total cholesterol
- Lower HDL
- Dyslipidemia less likely to be treated in ACHD

Diabetes/MetSyn

- Diabetes and metabolic syndrome increased in ACHD
- Highest in cyanotic groups, Turner syndrome, Down syndrome (DM1)

Tobacco

- Less common in ACHD
- More prevalent with more severe disease
- Increased mortality among smokers

Brida et al. Eur Heart J 2023

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#5 A WORD ON ARRHYTHMIAS

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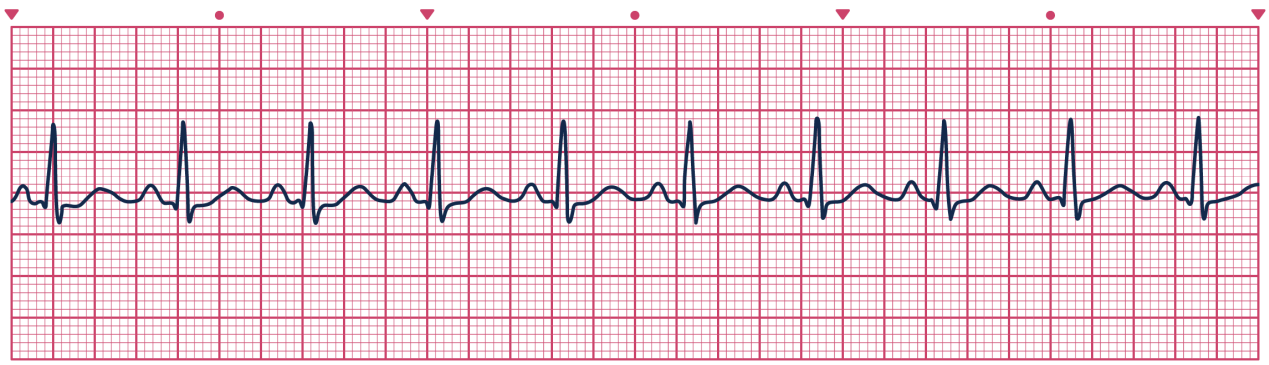
RHYTHM CONTROL, NOT JUST RATE CONTROL



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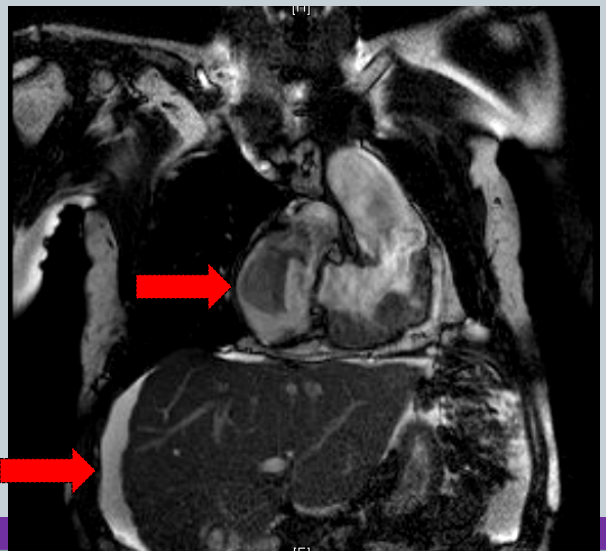
Tricuspid atresia with Fontan repair and atrial fibrillation



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#6 WHAT MAKES ACHD 'DIFFERENT'?

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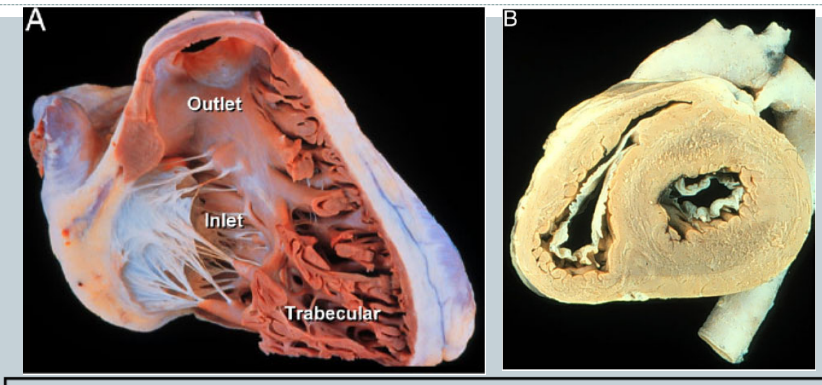
Right ventricles are not left ventricles



Relies on longitudinal shortening

Usually pumps to a low resistance circuit

Right coronary supplies RV

Warnes, *J Amer Coll Cardiol* 2009

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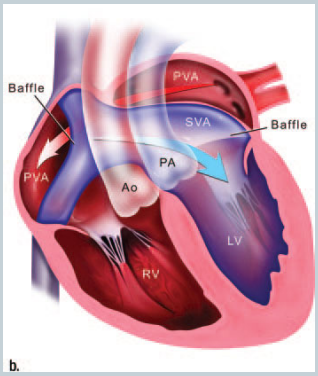
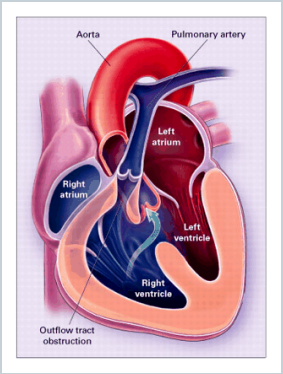
ITERATIONS OF THE RIGHT VENTRICLE
AKA: "WHY ACHD CAN BE SO HARD"

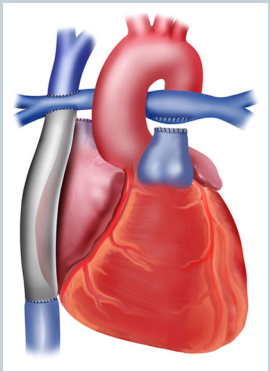
SYSTEMIC RV
TRANSPOSITION



SUBPULMONIC RV
TETRALOGY OF FALLOT



SINGLE RV
HYPOPLASTIC LV



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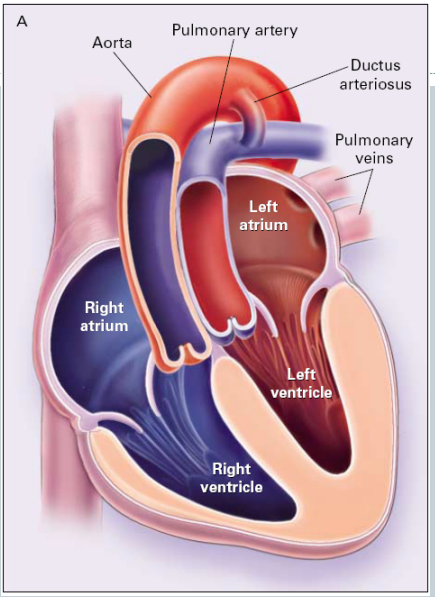

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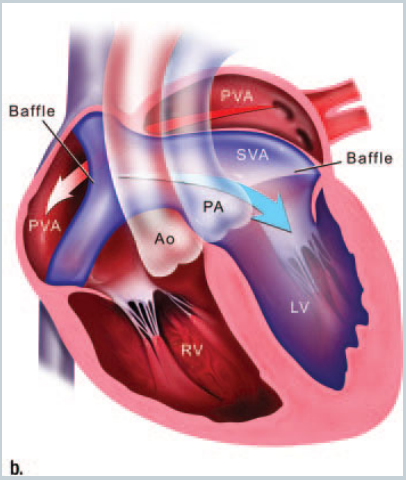
TRANSPOSITION OF THE GREAT ARTERIES
COMPLICATIONS OF ATRIAL SWITCH

- **RV dysfunction**
  may be due to relative ischemia of hypertrophied RV fed by RCA
- **Baffle obstruction in 5-10%**
  usually superior baffle
- **Baffle leaks in up to 25%**
  rarely require correction
  20 year survival 76 - 80%



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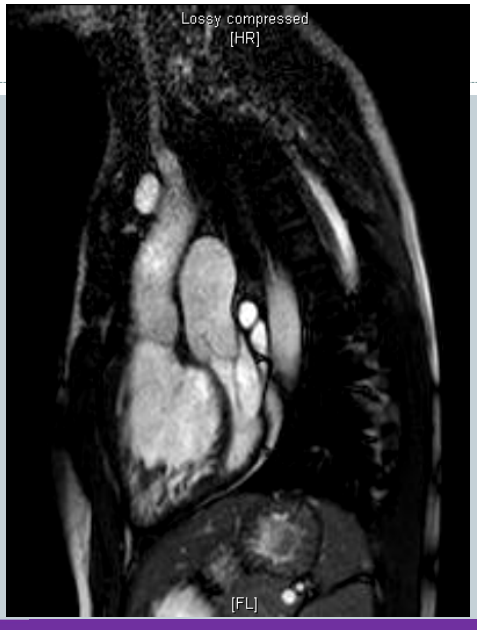

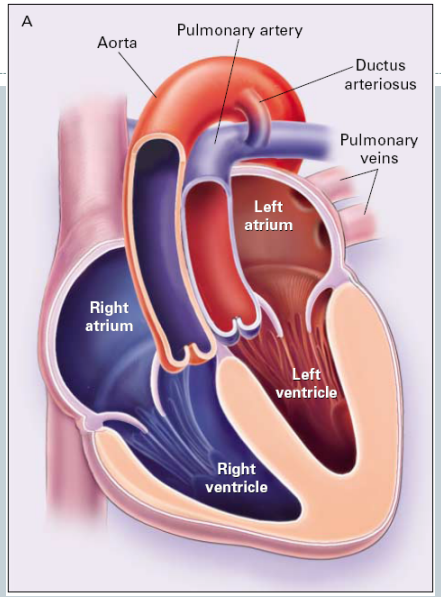

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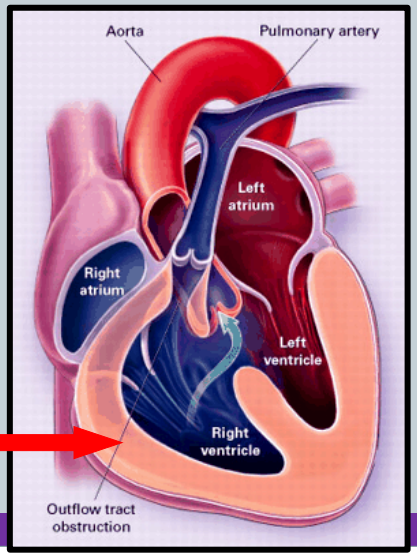
TETRALOGY OF FALLOT
ANATOMY

RIGHT VENTRICULAR OUTFLOW TRACT OBSTRUCTION

VENTRICULAR SEPTAL DEFECT

OVERRIDING AORTA

RIGHT VENTRICULAR HYPERTROPHY



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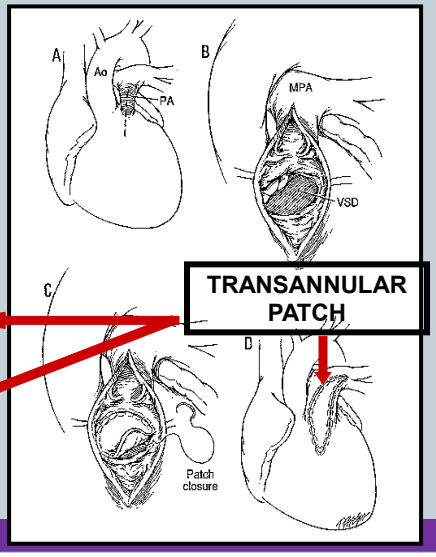
TETRALOGY OF FALLOT
SURGICAL REPAIR

- **Not cyanotic**
- **Normal pulmonary blood flow**
- **Pulmonary regurgitation**
- **Right ventricular enlargement**



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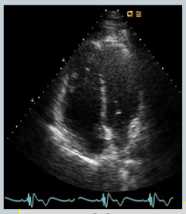
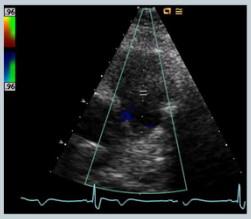
The adult with repaired tetralogy of Fallot
Spectrum of severity

FINE



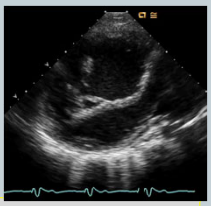
PRETTY GREAT

22 year old asymptomatic, normal right ventricular function, normal pulmonary valve



40 year old with abnormal exercise test, significant pulmonary regurgitation, large right ventricle that doesn't work normally

NOT AS GREAT



20 year old with branch pulmonary stenosis, atrial flutter, hospitalized several times a year for heart failure

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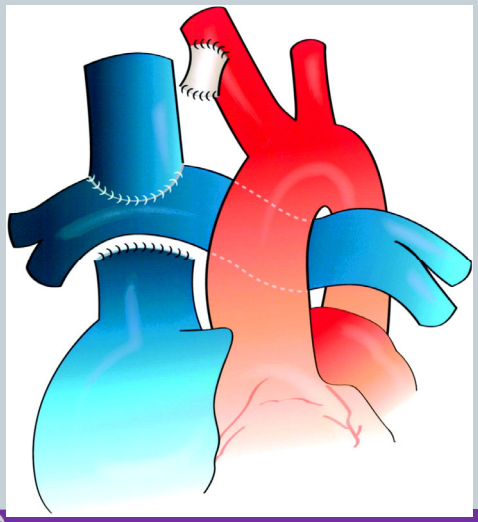
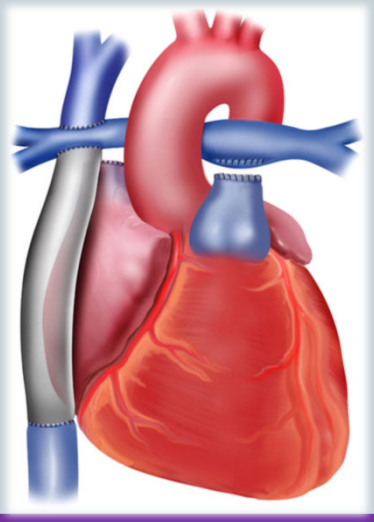
Glenn and extra cardiac Fontan





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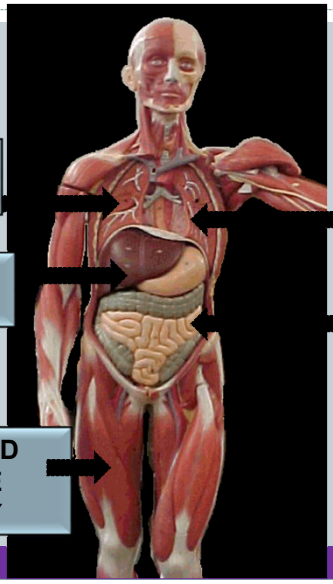
Fontan palliation is a multisystem disorder



PULMONARY EMBOLI

LIVER DYSFUNCTION

DECREASED EXERCISE CAPACITY

ARRHYTHMIAS

PROTEIN LOSING ENTEROPATHY

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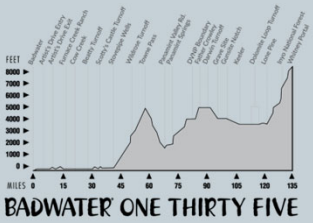
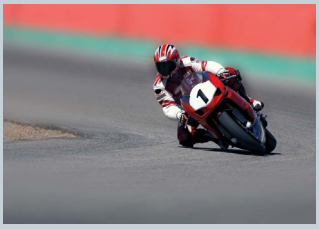
It's a younger population who may want to do things not typical of older patients with heart disease





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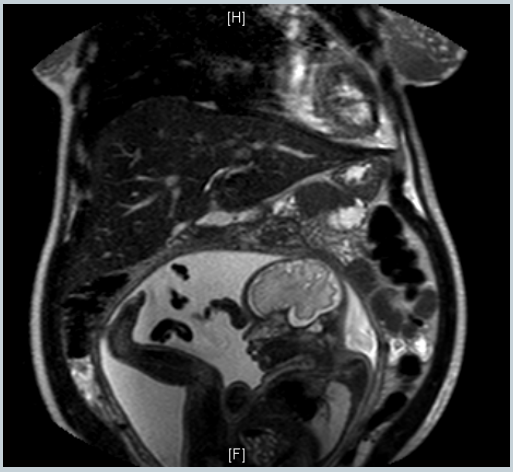
Pregnancy + ACHD

Pregnancy is a hemodynamic challenge

Most patients can do well

Challenges with more complex or symptomatic CHD



45

Remember the age of these patients: Surgical PVR over the lifetime of a patient with TOF

Initial repair → Age 15 years → 25 years → 40 years → 55 years → 70 years

6 or more sternotomies over a lifetime

46

#7 WHAT ABOUT HEART FAILURE IN ACHD

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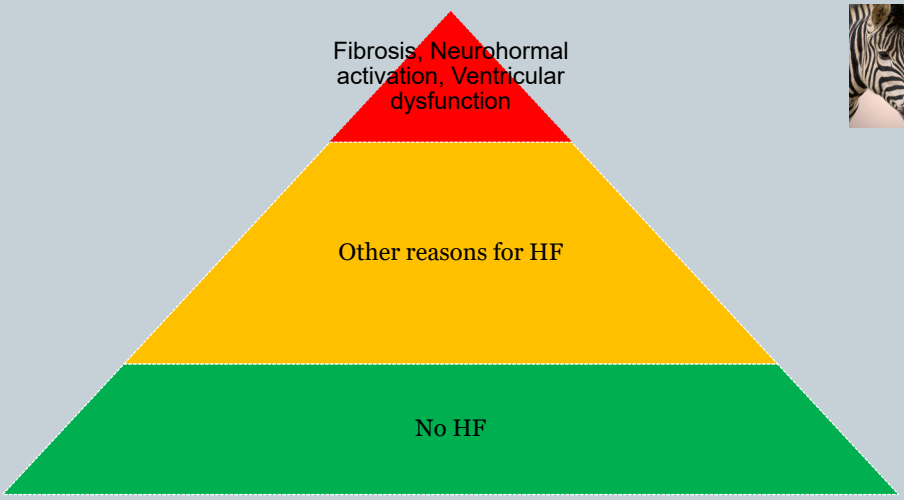
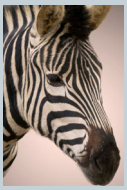
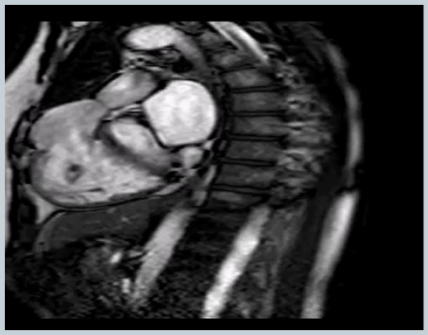
Some ACHD patients might benefit from 'standard' HF medical therapy

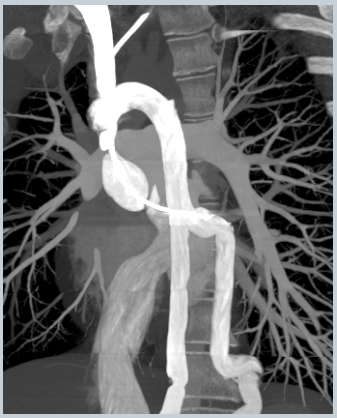


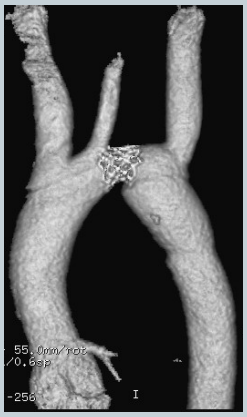
49

Some causes of "heart failure" are reversible









50

Simplistic look at the effect of heart failure medications in acquired HF

Reduce preload
Reduce afterload

Improve contractility
Minimize fibrosis

Many benefits are due to neurohormonal effects

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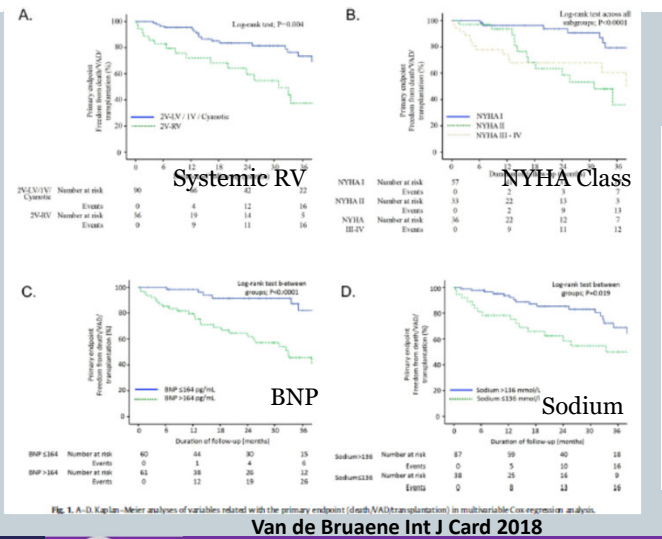
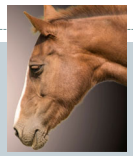
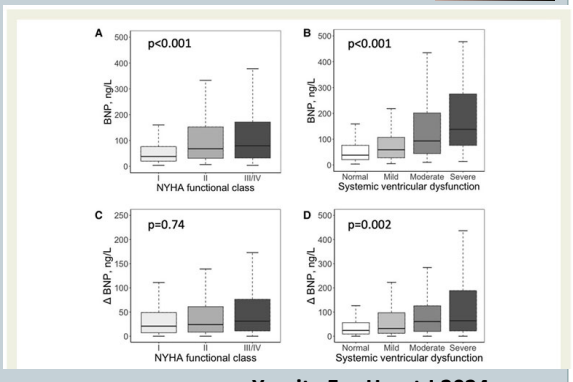
MARKERS OF RISK MAY BE SIMILAR TO ACQUIRED HF



Van de Bruaene Int J Card 2018





Yumita Eur Heart J 2024

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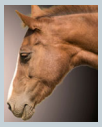
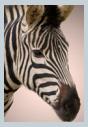
We don't have as much data

• LV SYSTOLIC DYSFUNCTION	• ACHD
• Large trials – 1000's of patients • Large numbers of significant events over short months to years • Relatively little heterogeneity in etiology • Many prognostic markers 	• Tiny trials – 10's of patients • Few events, often occurring over decades • Extremely heterogeneous anatomy, physiology, surgical history • Fewer prognostic markers 

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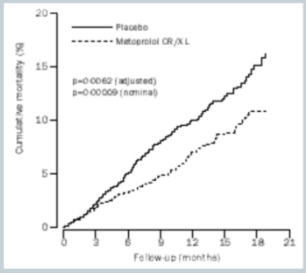
ACEI and Beta blockers in HF – early data

SOLVD

- 2579 patients
- NYHA Class II-III, EF<35%
- Double blind, randomized, placebo controlled trial of enalapril
- Avg follow-up ~ 41 months
- Deaths: 510 (39.7%) placebo vs 452 (35.2%) enalapril
- 26% risk reduction for hospitalization

MERIT- HF

- 3991 patients
- NYHA II-IV
- On ACE I/ARB
- EF<40%
- Mean f/u 1 year



- Deaths:
- 7.2% (Toprol XL) vs. 11% (placebo)

SOLVD Investigators *NEJM* 1991 MERIT-HF Study Group *Lancet* 1999

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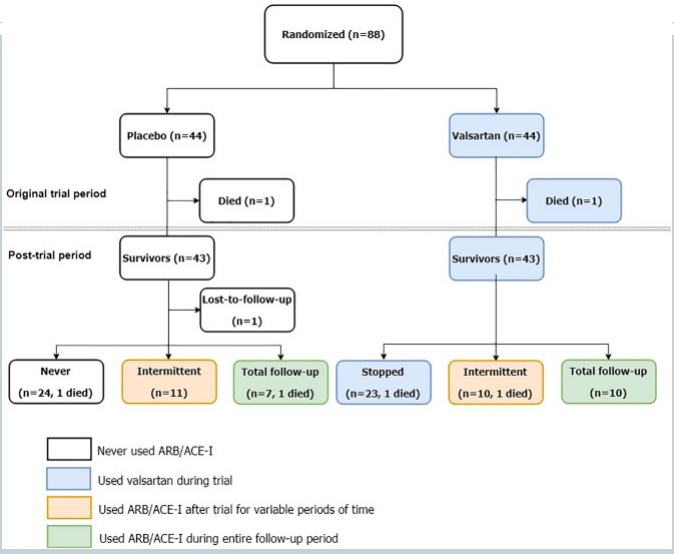
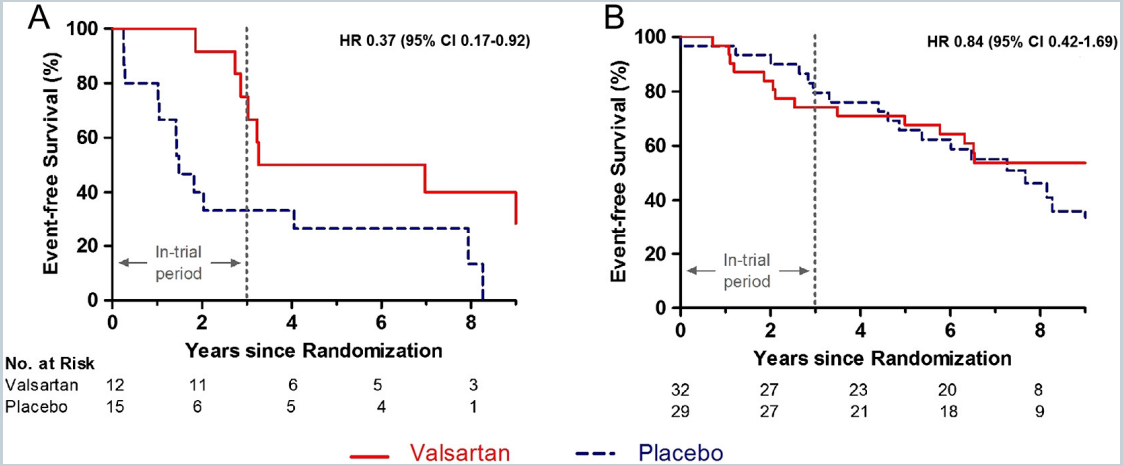
ARB/ACEI in Systemic Right Ventricles



55

Symptoms matter

Van Dissel Int J Card 2019



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Sacubitril/valsartan

Original Article

Early Experience With Sacubitril/Valsartan in Adult Patients With Congenital Heart Disease

Gentian Lluri, MD, PhD[1], Jeannette Lin, MD[1], Leigh Reardon, MD[1], Pamela Miner, RN, MN, NP[1], Katrina Whalen, RN, MSN, NP[1], and Jamil Aboulhosn, MD[1]

World Journal for Pediatric and Congenital Heart Surgery
2019, Vol. 10(3) 292-295

DOI: 10.1177/2150135119825599
journals.sagepub.com/home/pch
SAGE

ORIGINAL ARTICLE

Heart, Lung and Circulation (2020) 29, 137–141
1443-9506/04/$36.00
https://doi.org/10.1016/j.hlc.2018.12.003

Sacubitril/Valsartan in Adult Congenital Heart Disease Patients With Chronic Heart Failure – A Single Centre Case Series and Call for an International Registry

Vinesh Appadurai, MBBS [a,b,c*], Jennifer Thoreau, MBBS [a,b], Theresa Malpas, BSN [d], Mugur Nicolae, MD FRACP [a,c,d]

Original research

Sacubitril/valsartan in the treatment of systemic right ventricular failure

Tjitske E Zandstra [1,2], Marieke Nederend,[1] Monique R M Jongbloed [1,2], Philippine Kiès,[1] Hubert W Vliegen,[1] Berto J Bouma,[3] Laurens F Tops,[4] Martin J Schalij,[4] Anastasia D Egorova [1]

Sacubitril/valsartan for heart failure in adults with complex congenital heart disease

Susanne J. Maurer [1], Claudia Pujol Salvador [1], Sandra Schiele [1], Alfred Hager [1], Peter Ewert [1], Oktay Tutarel [*,1]
Department of Congenital Heart Disease and Paediatric Cardiology, German Heart Centre Munich, Technical University of Munich, Munich, Germany

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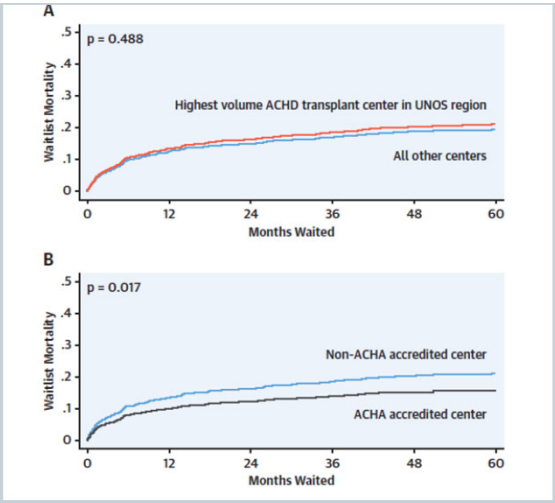
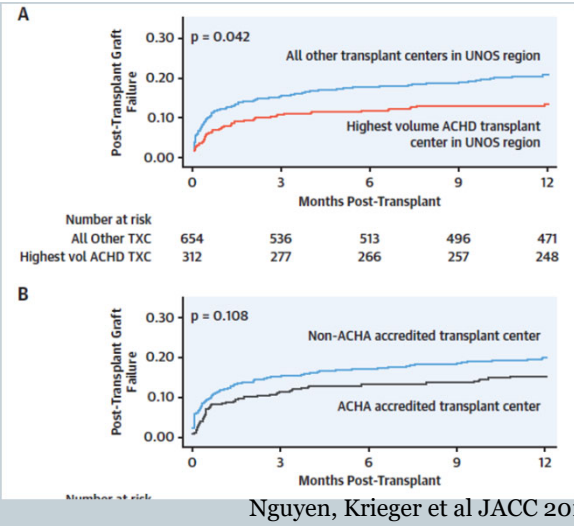
TRANSPLANT MORTALITY IN ACHD





Nguyen, Krieger et al JACC 2019

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#7 HOW SHOULD WE APPROACH HEART FAILURE IN ACHD?

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"Frankly, I mess around until they feel better and their BNP decreases without destroying their kidneys.

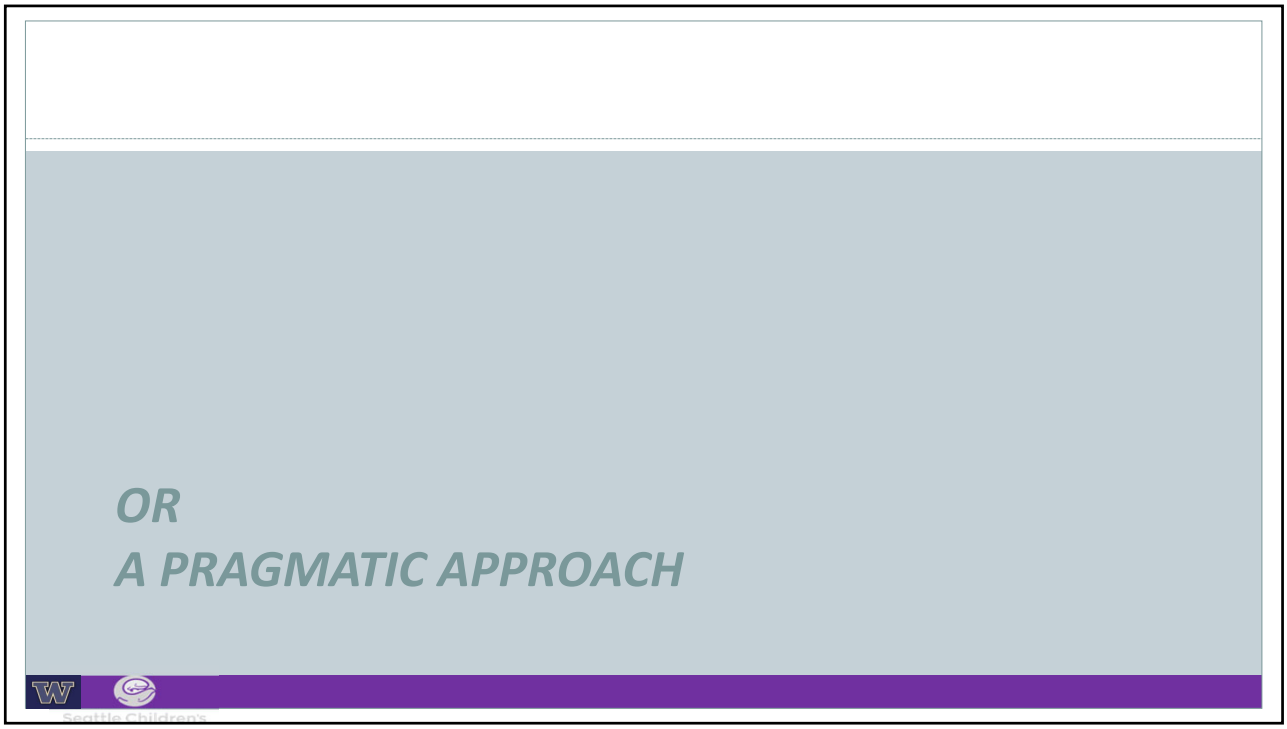
I then congratulate myself on my foresight and clinical acumen,

until they get overloaded again at which point I blame the nephrologist."

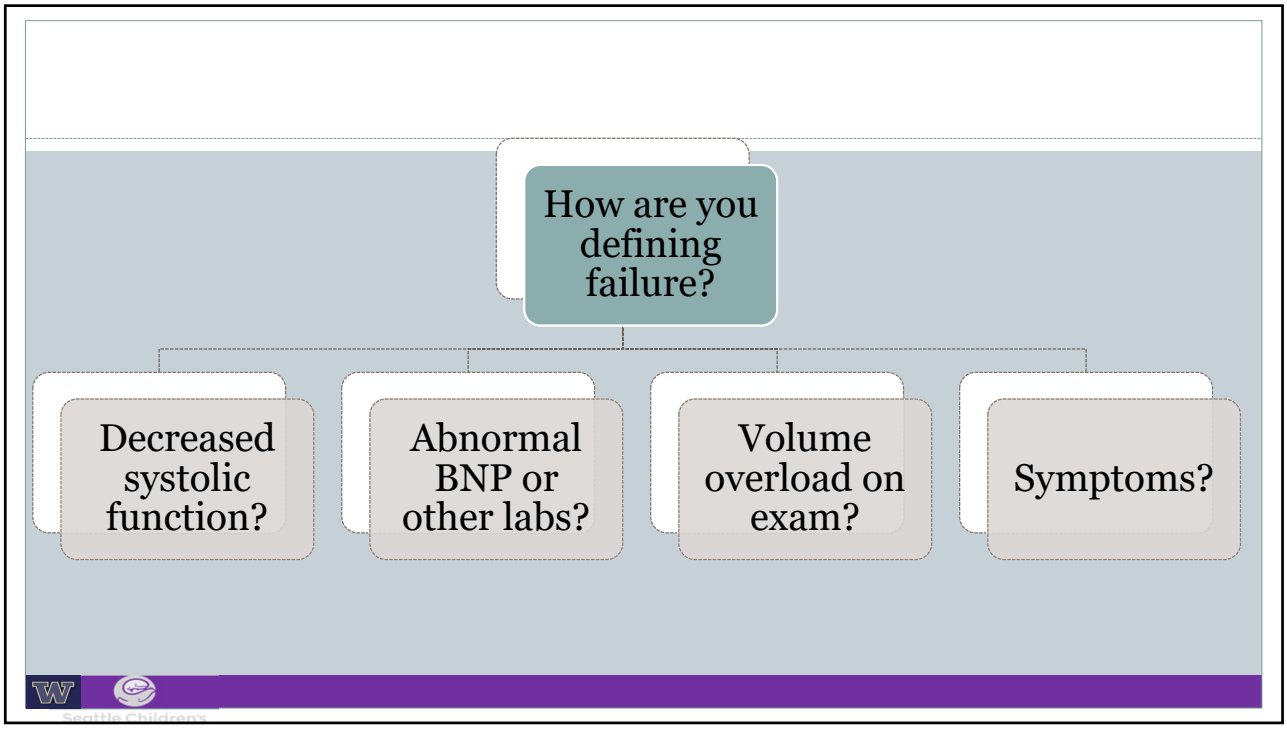
-Esteemed ACHD physician

ONE OPTION

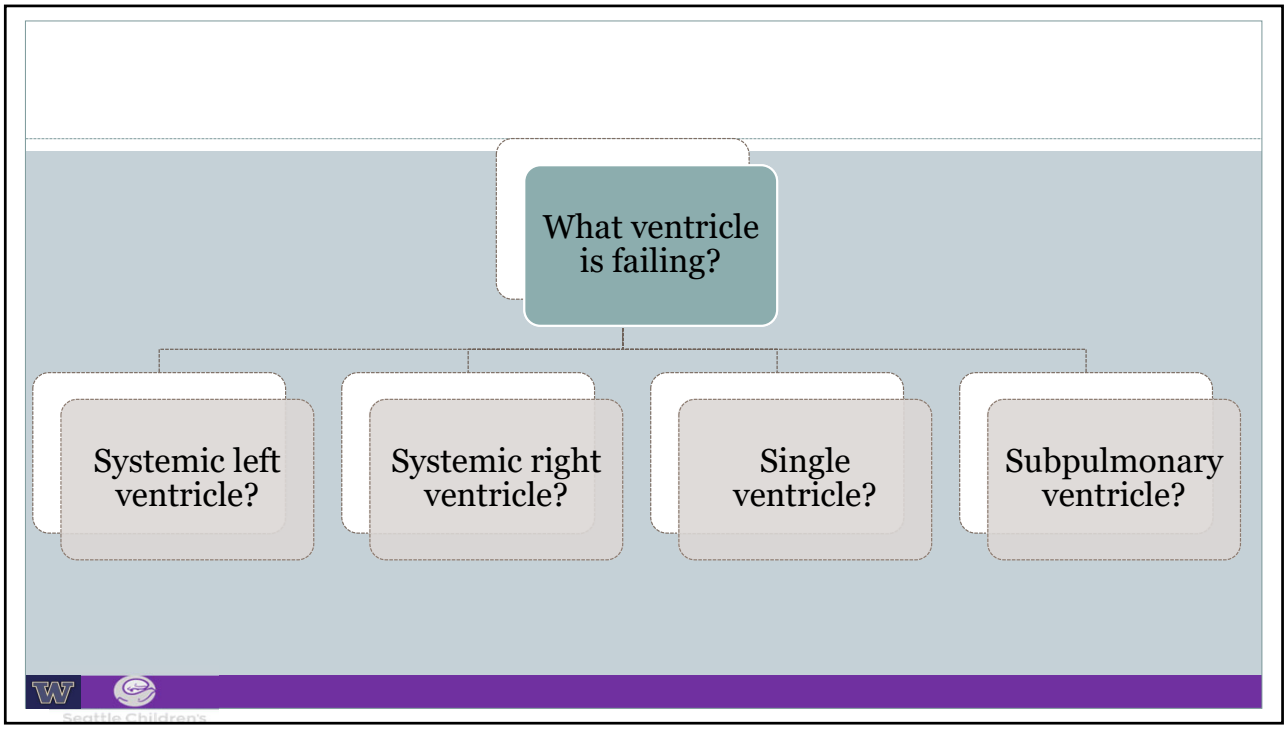
60


OR
A PRAGMATIC APPROACH

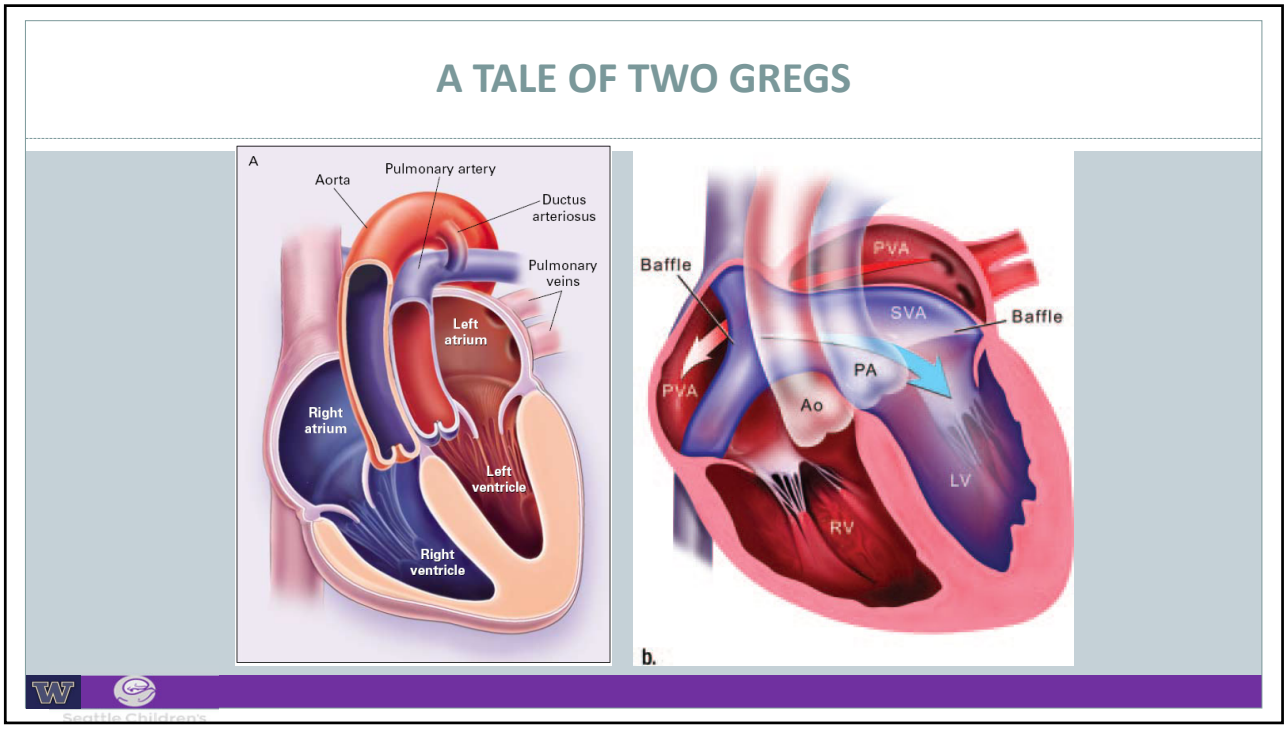
61


How are you defining failure?
Decreased systolic function?
Abnormal BNP or other labs?
Volume overload on exam?
Symptoms?

62


What ventricle is failing?
Systemic left ventricle?
Systemic right ventricle?
Single ventricle?
Subpulmonary ventricle?

63


A TALE OF TWO GREGS
A
Aorta
Pulmonary artery
Ductus arteriosus
Pulmonary veins
Left atrium
Right atrium
Left ventricle
Right ventricle
Baffle
PVA
SVA
Baffle
PA
PVA
Ao
LV
RV
b.

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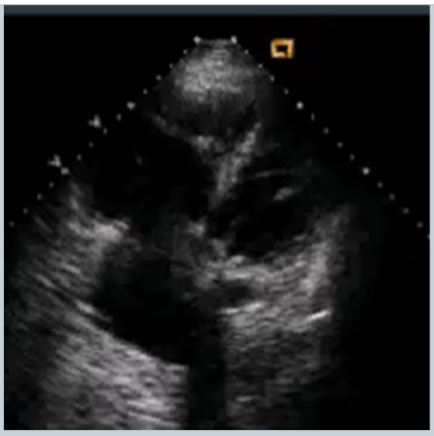
A TALE OF TWO DOUGS

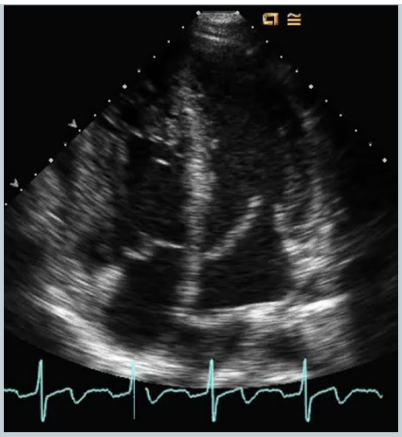
GREG #1	GREG #2
43 year old man	**35 year old man**
• **NYHA II-IV symptoms**	• **Asymptomatic**
• **ICD for atrial and ventricular arrhythmias**	• **Bradycardia at rest**

65

A TALE OF TWO GREGS



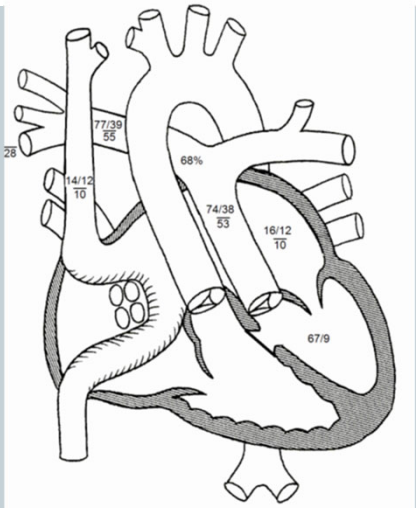
Greg 1



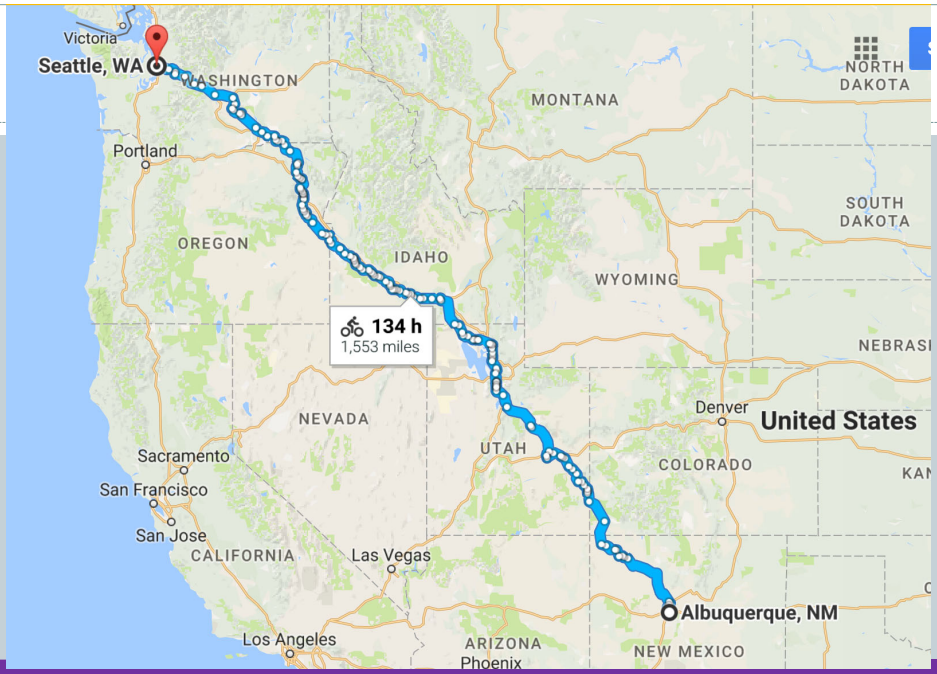
Greg 2

66

A TALE OF TWO GREGS



67



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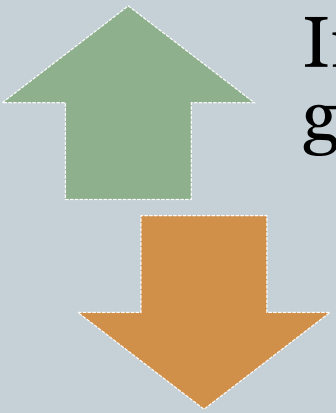
#8 MEDICATIONS AREN'T THE ONLY THING THAT CAN HELP

69

For ACHD patients with full time jobs – GRIT is magic sauce

Grit = passion and perseverance in pursuit of long-term goals



Increased grit

Decreased depression

Liu et al Applied Nursing Res 2024

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Brady

Works full time as an engineer
Married with three kids and a golden retriever
Plays tennis, active outdoors

In his own words:
Staying active (walking, exercising, sports)
Consistent and careful monitoring
Good mental health, putting less stress on my body and heart
Balanced diet
Great care and treatment from the beginning

Seattle Children's

71

#9 ACHD PROGRAMS ARE INVALUABLE FOR THE CARE OF ACHD PATIENTS

72

SEATTLE ADULT CONGENITAL HEART DISEASE PROGRAM

73

What do you need to build an ACHD program

- Expertise: ACHD cardiology, surgery, IC, EP, imaging
- Transition from pediatric cardiology
- Institutional investment
- A dedicated person(s) to spearhead the program
- Patients and families!

Barriers

Money

Expertise

Money

Time

Collaboration

Money

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#10 TAKING CARE OF ACHD PATIENTS IS AWESOME

75

Laura

- Initial palliation with BTT shunt, then lateral tunnel Fontan at age 4
- Teacher, married, 2 kids
- Regularly plays soccer
- Coiled APCs, PVCs, rare AT, mild cirrhosis
- VO2 max 75% predicted

76

30 year old with a history of CHD

77

conclusions

- ACHD is increasingly common
- There aren't enough ACHD experts
- It takes a village of providers to outcomes
- Most patients do well, some problems are preventable
- ACHD programs are valuable, and need to be 'hub and spoke'
- Call your friendly ACHD doc for help!

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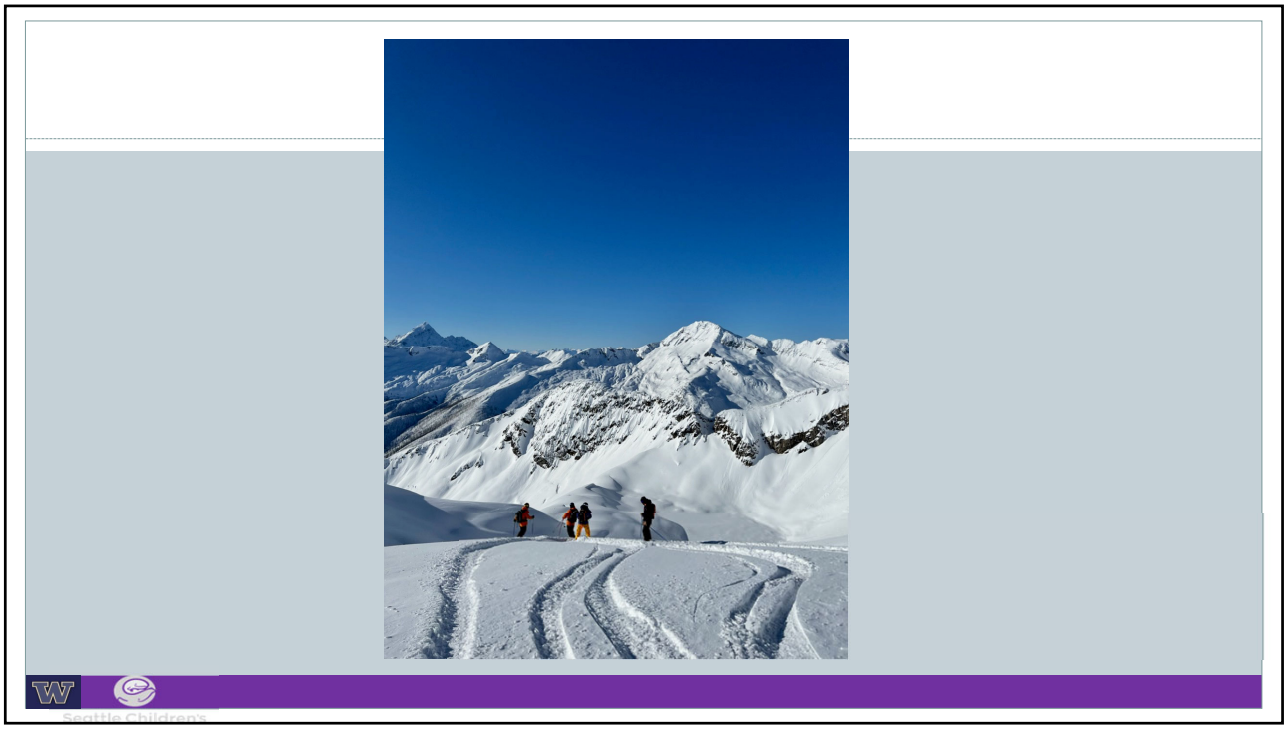

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