

CASE 1: 24 weeks

24W R/O CHD



24W R/O CHD

BREECH



SAMSUNG
HEA110

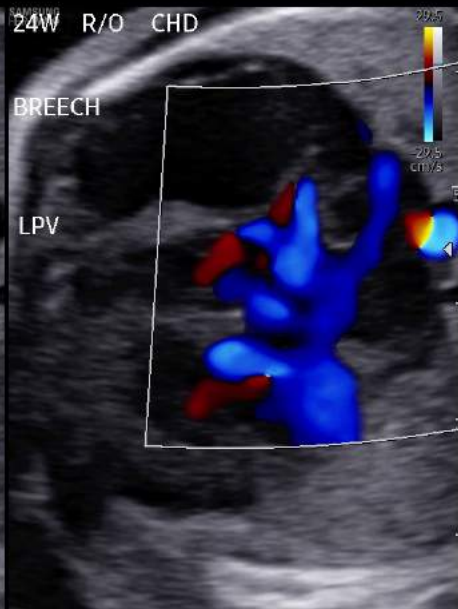


SAMSUNG

24W R/O CHD

BREECH

LPV



24W R/O CHD

BREECH

Ao_Arch



29 weeks

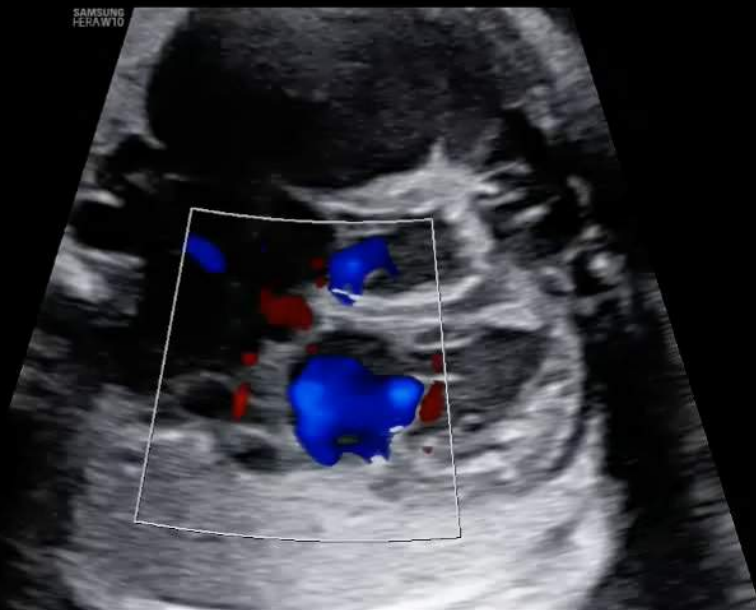


SAMSUNG
HERA W10



100

SAMSUNG
HEA W10



53.1

-53.1
cm/s

AoV

SAMSUNG
HEAWTO



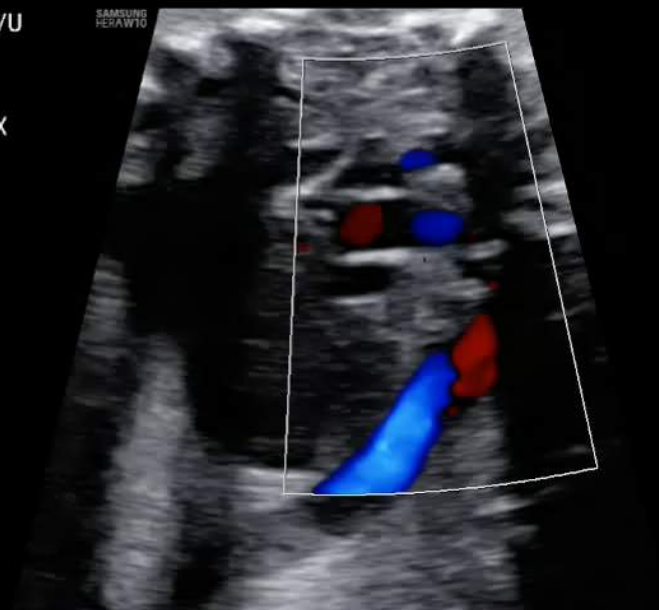
51

29W6 F/U

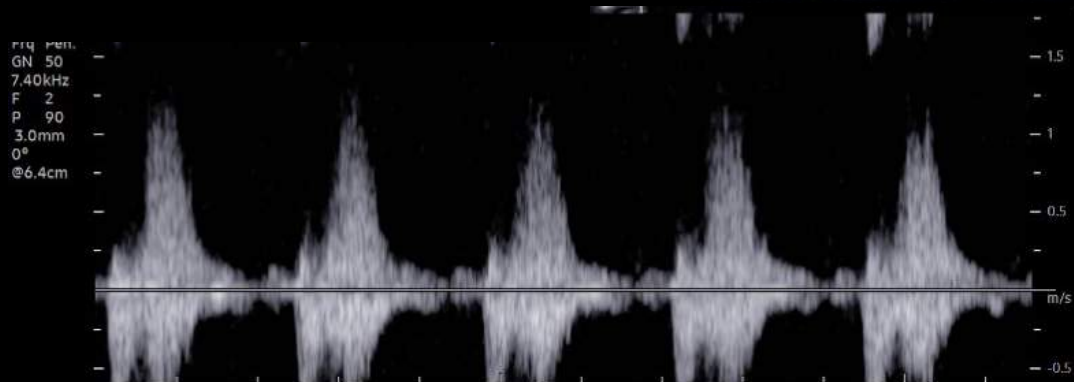
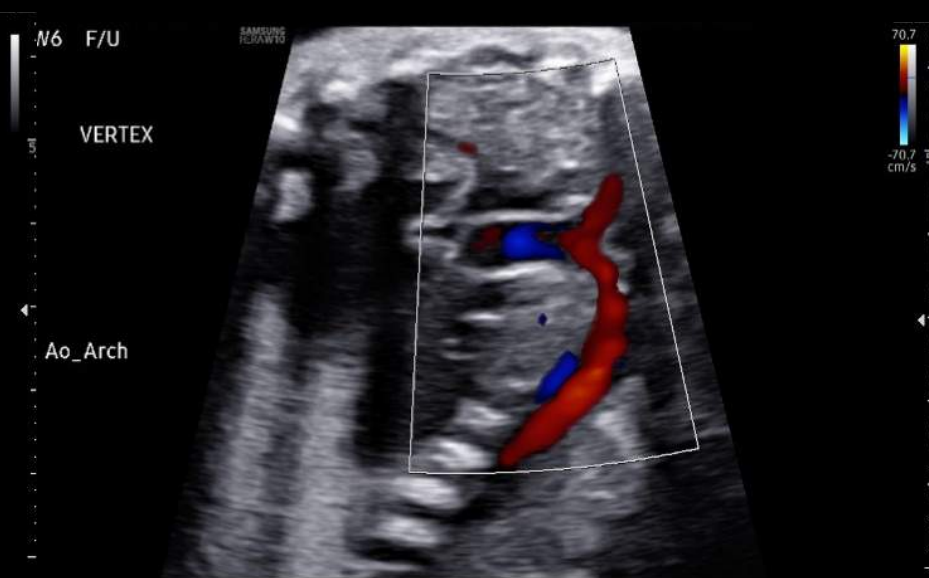
SAMSUNG
HEA110

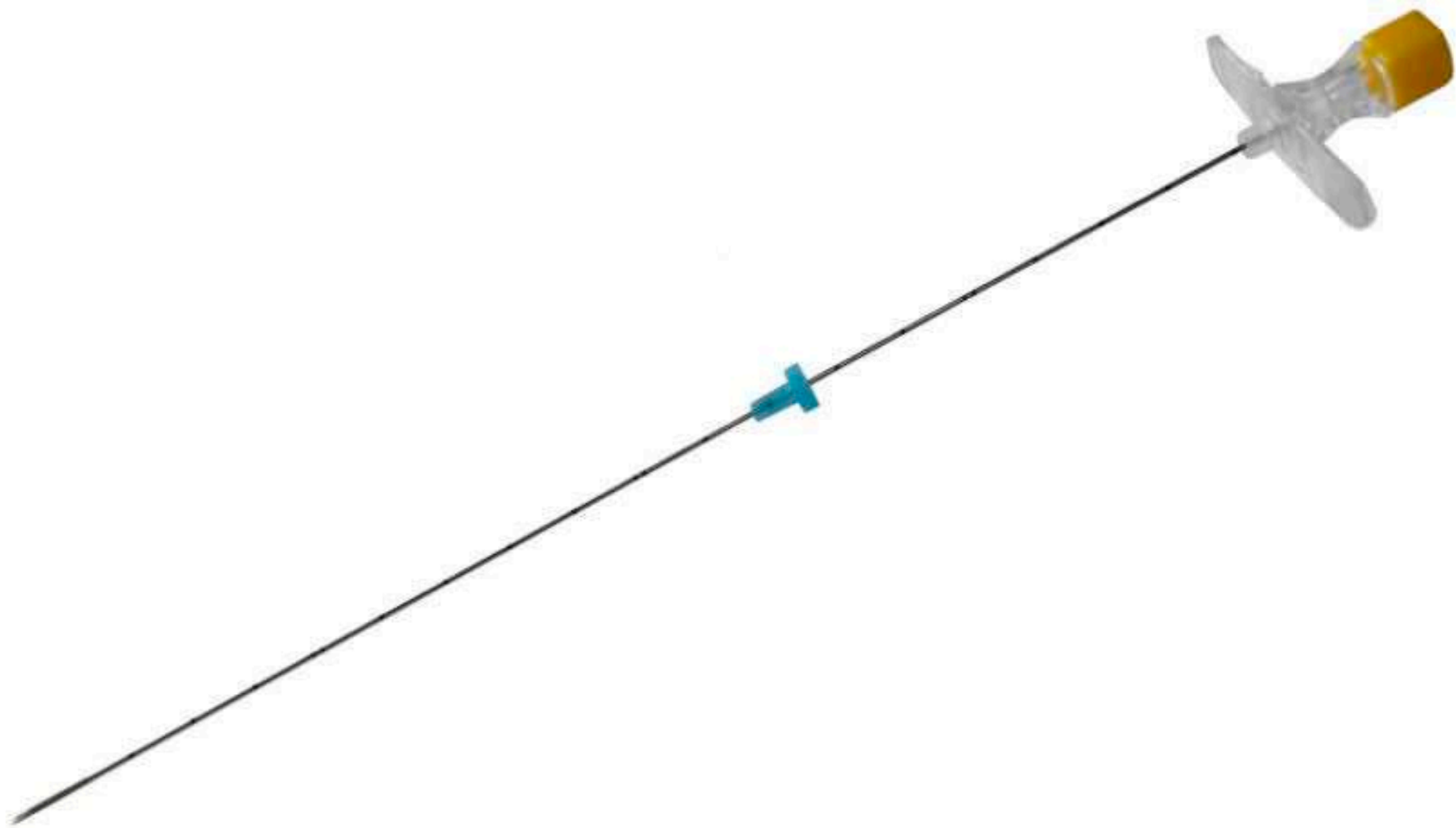
VERTEX

Ao_Arch



70.7
-70.7
cm/s





SAMSUNG
HERA W10







Postnatal ECHO

CHOP S9
S9-2
91Hz
7.0cm
2D
70%
C 48
P Low
HGen

TIS1.9 MI 1.1

M3

G
P R
3.0 6.0



156 bpm

CHOP S9
S9-2
35Hz
7.0cm

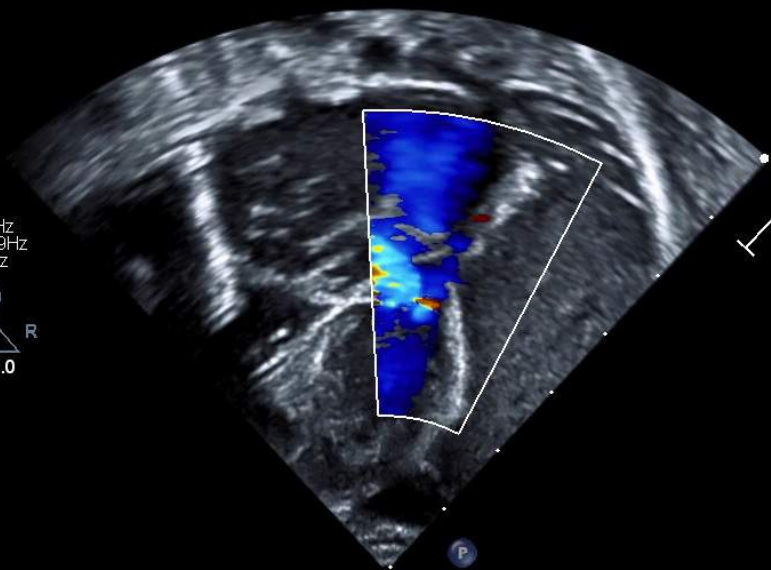
2D
71%
C 48
P Low
HGen

CF
40%
4300Hz
WF 429Hz
3.3MHz



TIS1.9 MI 1.0

M3 M4
+50.2
-50.2
cm/s



156 bpm

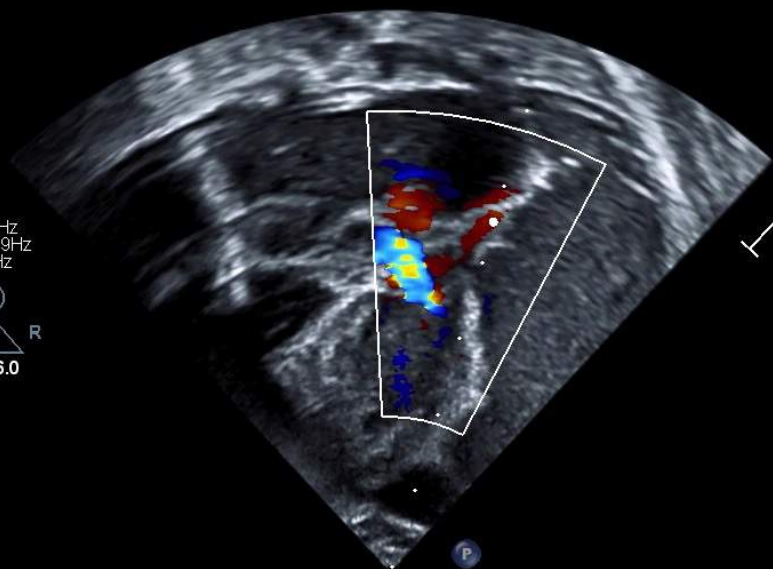
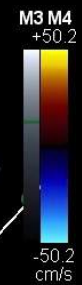
CHOP S9
S9-2
35Hz
7.0cm

2D
71%
C 48
P Low
HGen

CF
40%
4300Hz
WF 429Hz
3.3MHz



TIS1.9 MI 1.0

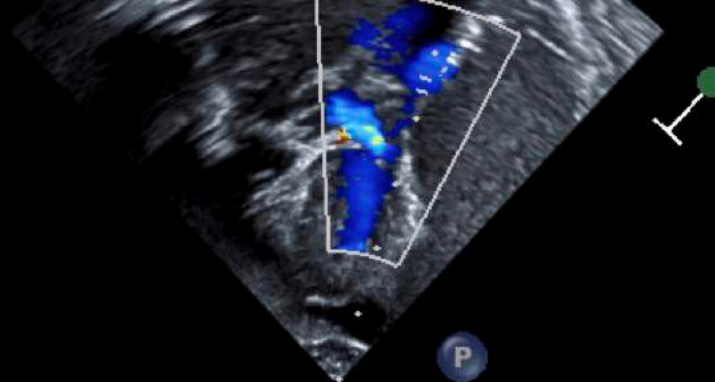


157 bpm

2D
70%
C 48
P Low
HGen

CF
40%
4300Hz
WF 429Hz
3.3MHz

PW
40%
WF 150Hz
SV2.0mm
3.0MHz
4.5cm



-50.2
cm/s

-80

-40

cm/s

-40

00

CHOP S9
S9-2
91Hz
7.0cm

2D
70%
C 48
P Low
HGen

TIS1.9 MI 1.1

M3



155 bpm



Denise Adams, MD is a pediatric hematologist-oncologist and Director of the Comprehensive Vascular Anomalies Program (CVAP) at Children's Hospital of Philadelphia. She holds the Alan R. Cohen Endowed Chair in Pediatrics.



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Blue Rubber Bleb Nevus Syndrome (BRBNS)



Contact Us

Contact the Vascular Anomalies Center
617-355-5226

Fax
617-730-0752

International
+1-617-355-5209

Email
vascular@childrens.harvard.edu

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What is blue rubber bleb nevus syndrome?

Blue rubber bleb nevus syndrome (BRBNS), sometimes called Bean syndrome, is a rare congenital vascular anomaly in which malformed veins, or blebs, appear on the skin and surfaces of internal organs. These small, purple lesions are particularly common in the gastrointestinal (GI) tract.

While a child with BRBNS can have hundreds of blebs on the skin, clinicians are generally more concerned with GI blebs, as they can bleed and cause anemia requiring iron supplements and blood transfusions.

There is no single accepted treatment for blue rubber bleb nevus syndrome. Both GI and skin blebs can be treated with surgery or sclerotherapy. However, doctors typically leave blebs on the skin alone unless they cause cosmetic problems, pain or, if located on the soles of the feet, difficulty walking.

Blebs can appear and grow throughout a child's lifetime, but those that are removed do not return.

How we care for blue rubber bleb nevus syndrome

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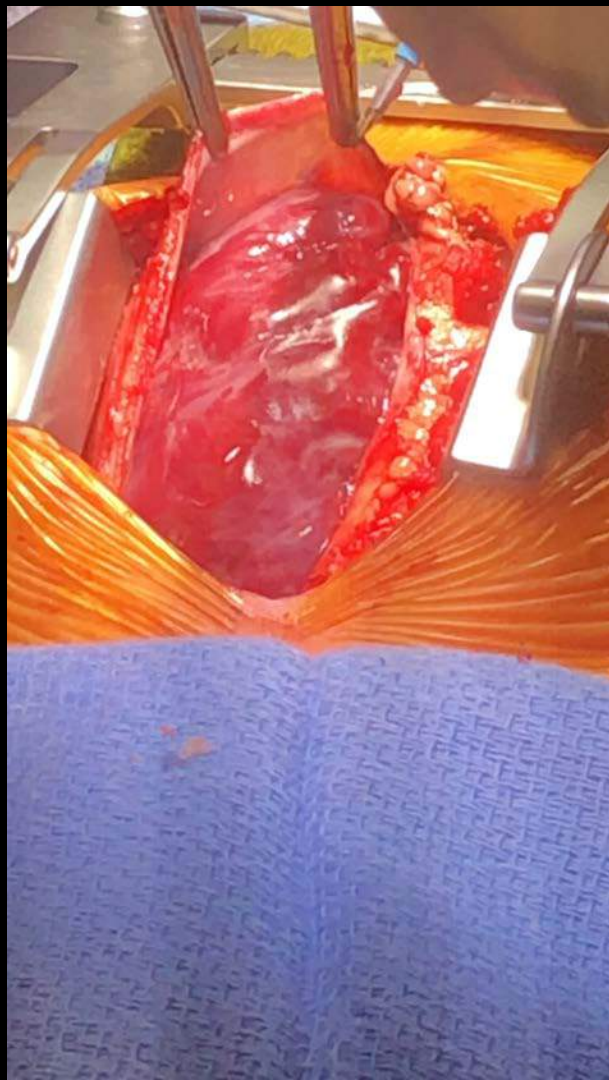
Steven J. Fishman MD

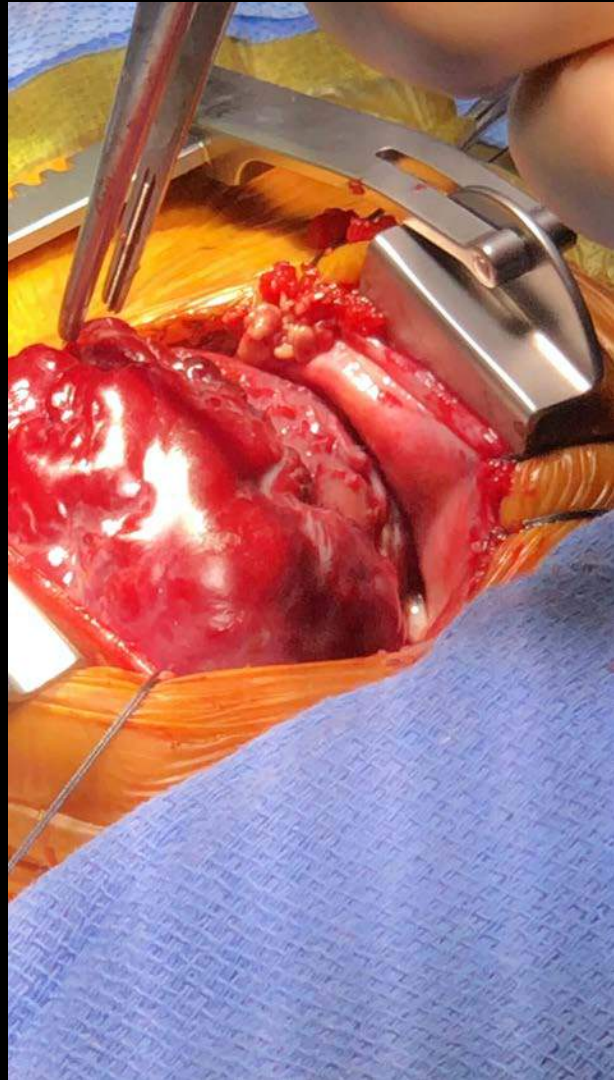
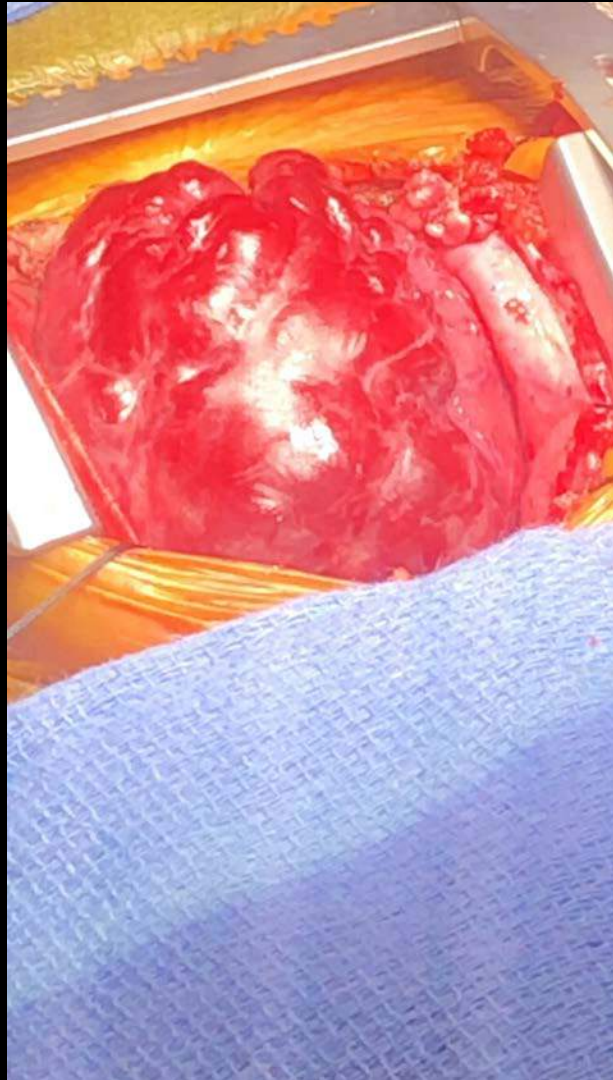
Surgeon-in-Chief; Chief, Department of Surgery; Stuart and Jane Weitzman Family Chair in Surgery; Co-Director, Vascular Anomalies Center

Professor of Surgery, Harvard Medical School

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Gender: Male

Test Requested: Somatic Overgrowth & Vascular Malformations Panel (v3)

Sample(s) Analyzed:

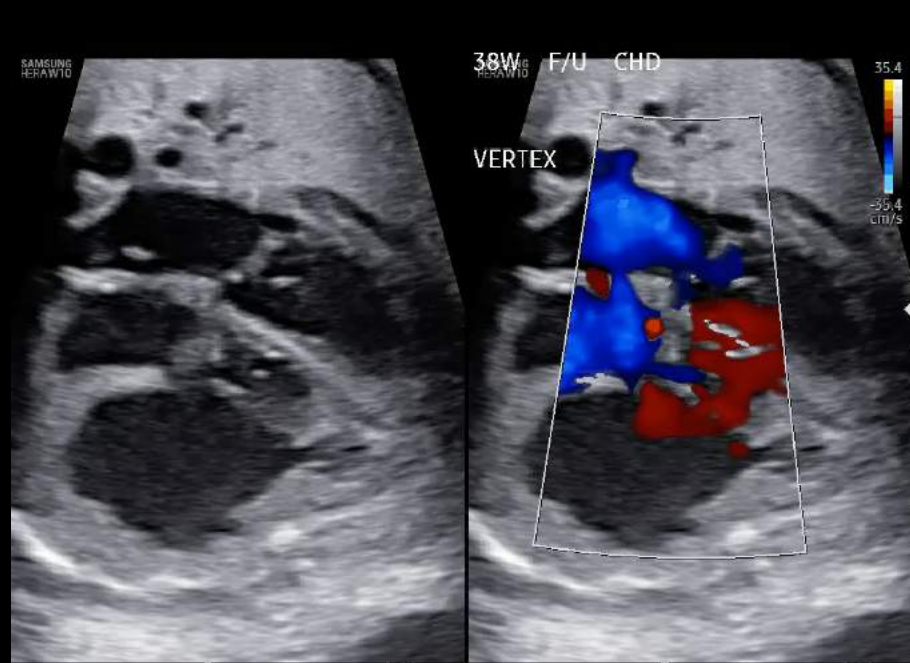
- 1: Genomic DNA isolated from blood
- 2: Genomic DNA isolated from Left foot frozen tissue sample
- 3: Genomic DNA isolated from Left foot FFPE sample

Result Summary: POSITIVE Two Somatic Pathogenic Variants in *TEK*

Identification of Variant(s):

<u>Sample</u>	<u>Gene</u>	<u>Nucleotide Position</u>	<u>Amino Acid Position</u>	<u>Variant Frequency</u>	<u>Classification</u>
2	<i>TEK</i>	c.2690A>T	p.Tyr897Phe	10.3 - 12.5%	Pathogenic
2	<i>TEK</i>	c.2753G>A	p.Arg918His	9.9 - 13.7%	Pathogenic

CASE 2: Fetus @ 38 weeks



CHOP S8
S8-3
87Hz
7.0cm

2D
68%
C 48
P Off
HGen



TIS1.1 MI 1.2 CHOP S8
S8-3
M4 44Hz
7.0cm

2D
59%
C 59
P Off
HGen

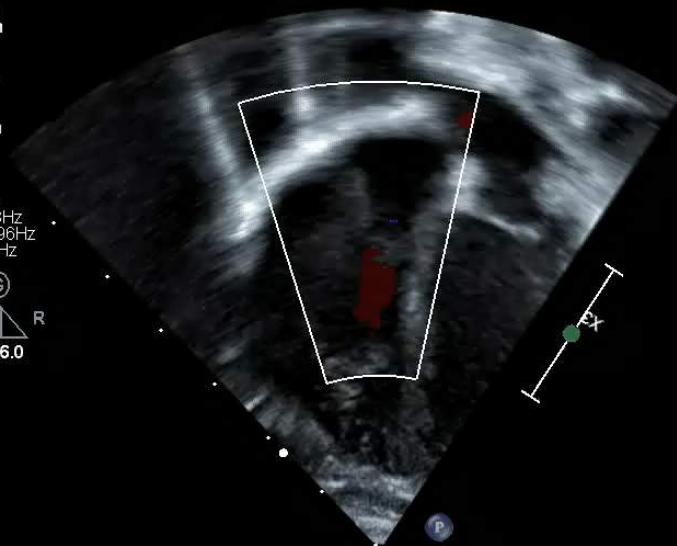
CF
40%
9968Hz
VWF 996Hz
3.3MHz



146 bpm

TIS1.6 MI 0.9

M4 M4
116



140 bpm

CHOP S8
S8-3
87Hz
7.0cm

2D
67%
C 48
P Off
HGen

P (G) R
3.0 6.0

TIS1.1 MI 1.2

M4

CHOP S8
S8-3
106Hz
6.0cm

2D
59%
C 59
P Off
HGen

P (G) R
3.0 6.0

TIS1.0 MI 1.0

M4

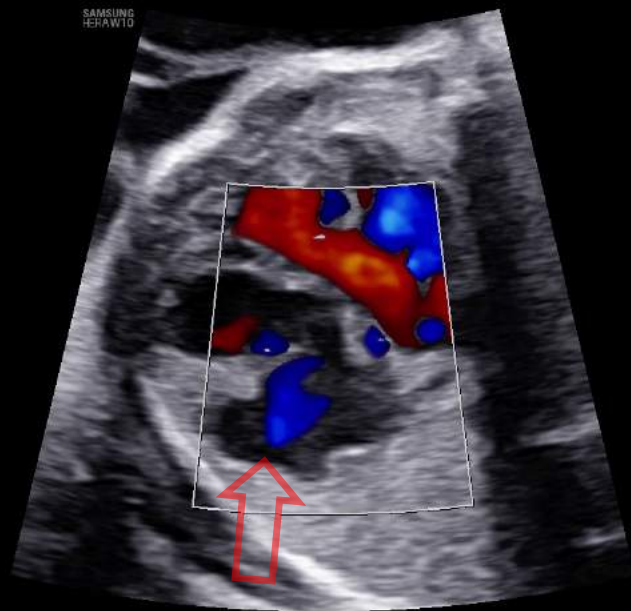
147 bpm

153 bpm

CASE 3



SAMSUNG
HEHAW10



53.1
-53.1
cm/s

10



SAMSUNG
RHAW10

10

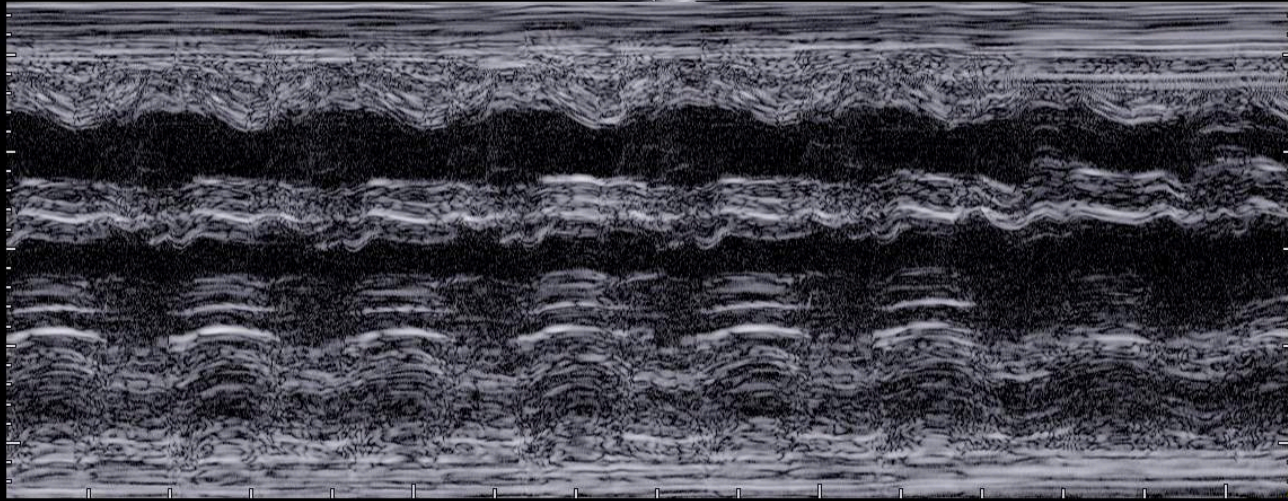
[2D]

Frq Gen.
GN 52
DR 48
FA 3
P 90

[M]

GN 50
DR 48
P 90

SAMSUNG
HERAW10



CASE 4: Presented @ 26 weeks



26W2D

SAMSUNG
HEAW10

VERTEX



26W2D

SAMSUNG
HEAW10

VERTEX



Back @ 29 weeks...



CHOP Cardiac C9 New
C9-2
55Hz
S1

GA 30w1d

TIB0.5 MI 0.9

2D
75%
Dyn R 48
P Med
HGen

Vertex



M2



12cm

*** bpm





CHOP Cardiac C9 New

C9-2

96Hz

S1

Z 1.2

2D

73%

Dyn R 48

P Med

HGen



11B0.9 MI 0.9

M2



X2

— *** bpm

CHOP Cardiac C9 New

C9-2

28Hz

Z 1.2

2D

84%

Dyn R 50

P Low

HGen

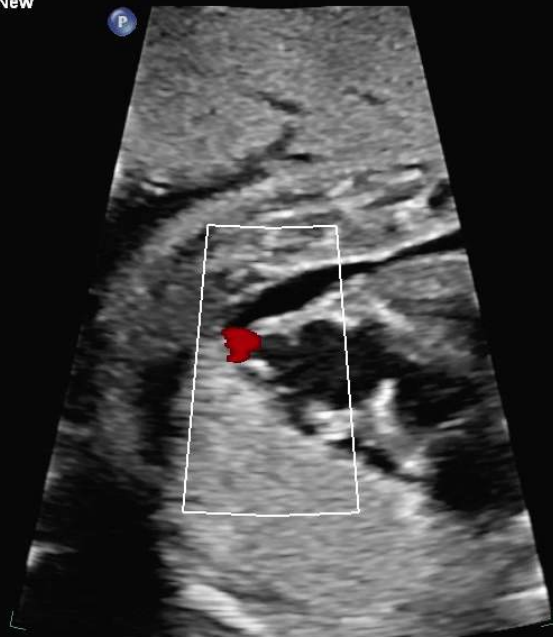
CF

48%

2850Hz

WF 185Hz

3.8MHz



11B1.0 MI 0.8

M2 M2

+28.9

2D

84%

Dyn R 50

P Low

HGen

CF

48%

2850Hz

WF 185Hz

3.8MHz



X2

— *** bpm

Back again @ 34 weeks





CHOP Cardiac C9 New
C9-2
63Hz
S1

GA 34w1d

TIB0.6 MI 0.9

2D
74%
Dyn R 48
P Med
HGen

M2



12cm

*** bpm

CHOP Cardiac C9 New

C9-2

76Hz

S1

2D

71%

Dyn R 48

P Med

HGen

TIB0.7 MI 0.9

M2



10cm

*** bpm

