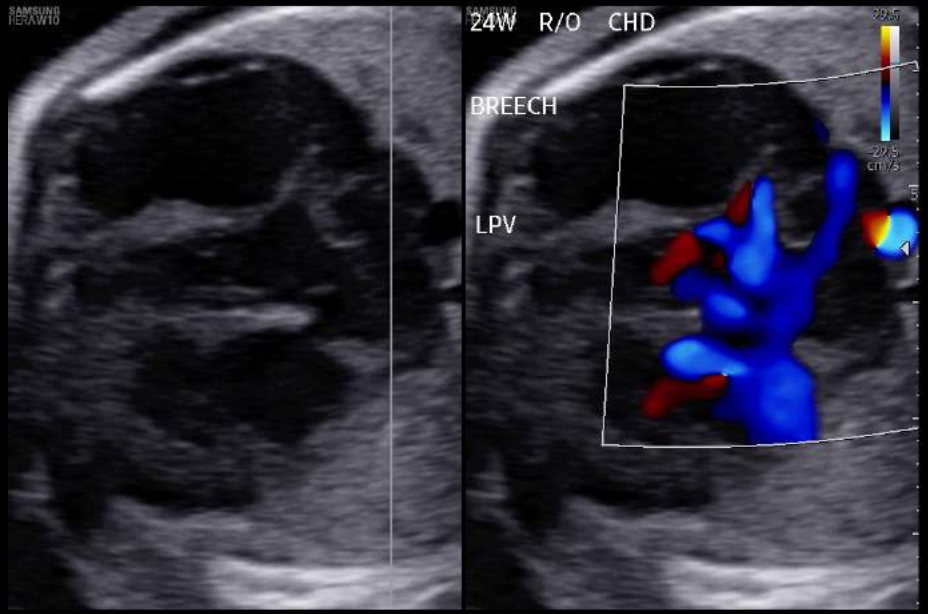
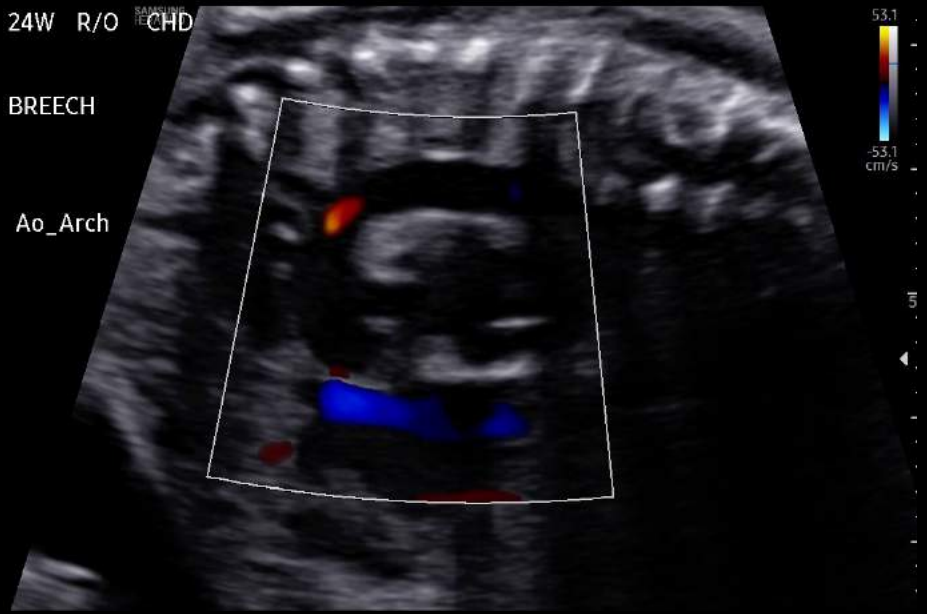
CASE 1: 24 weeks

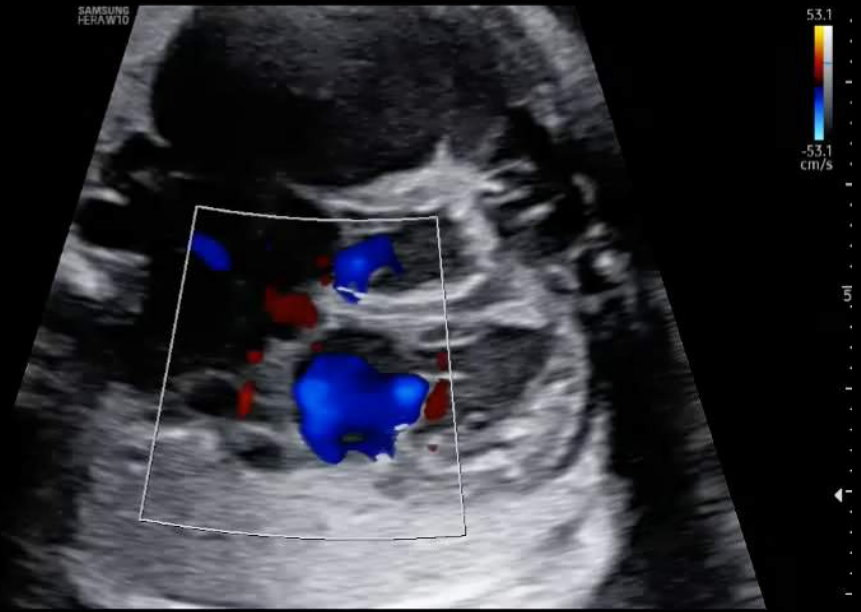

24W R/O CHD
BREECH
SAMSUNG
HS40


24W R/O CHD
BREECH
LPV


24W R/O CHD
BREECH
Ao_Arch
53.1
-53.1
cm/s

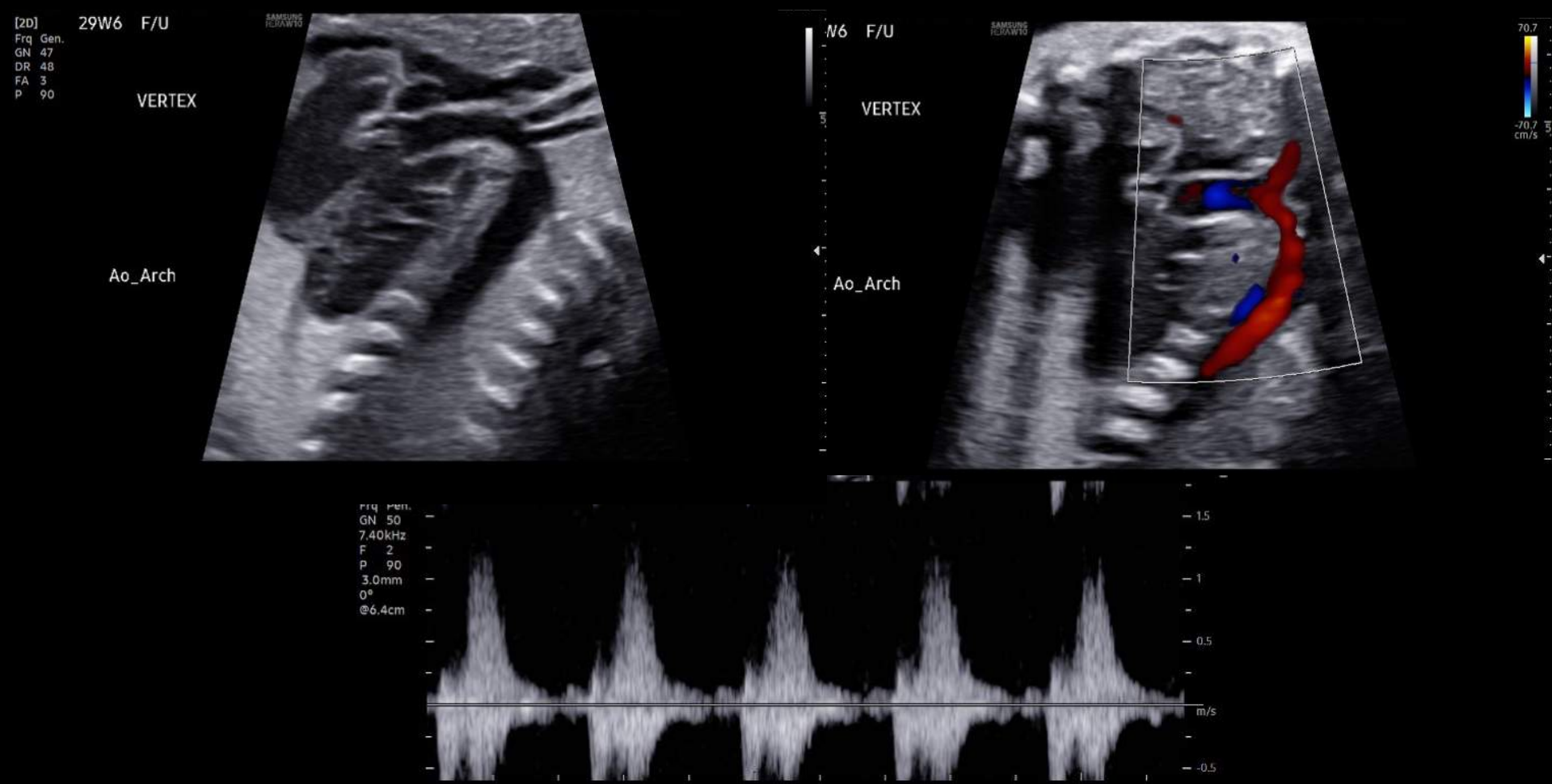
29 weeks

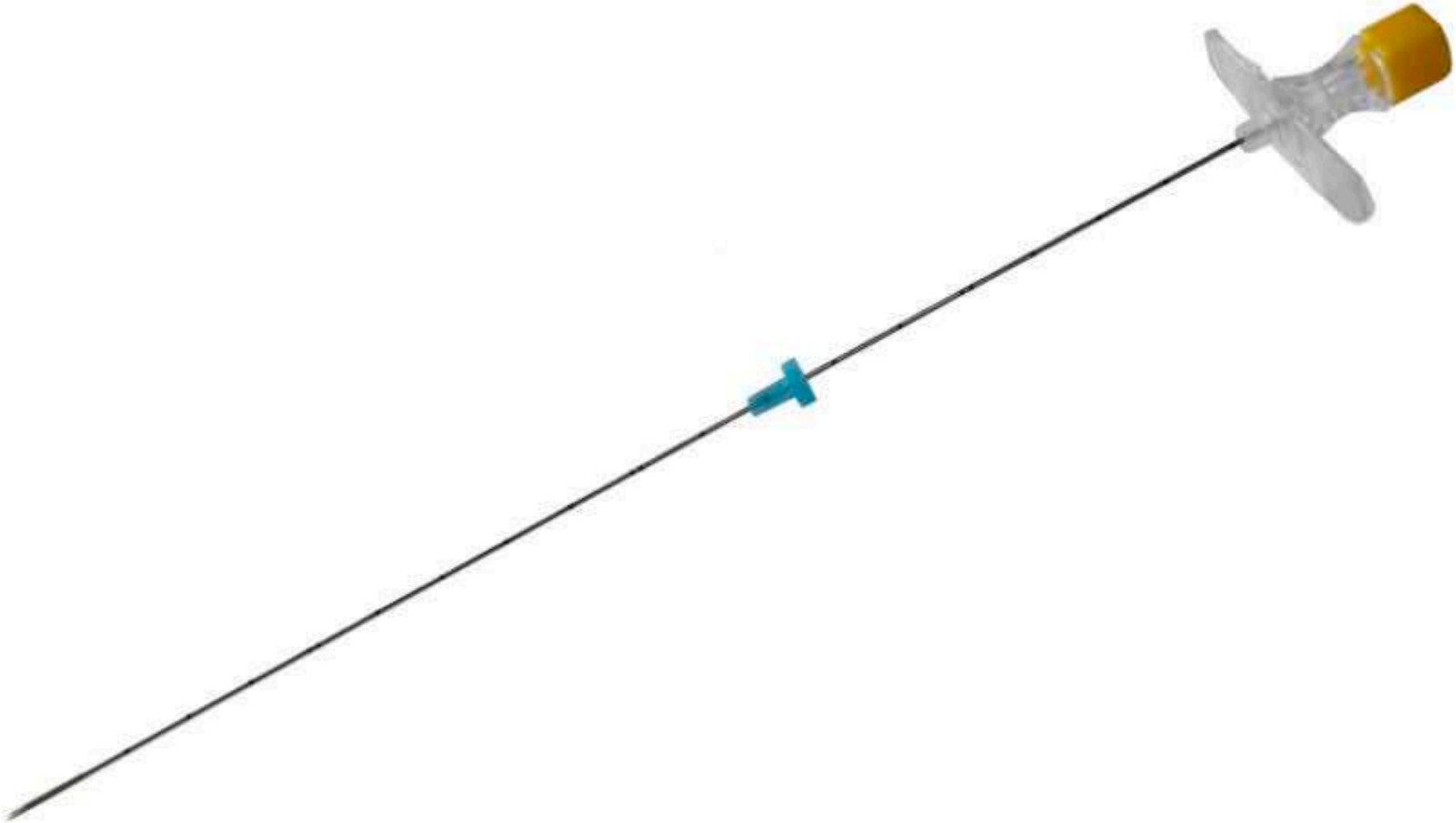

SAMSUNG
HERA W10


SAMSUNG
HERA W10
53.1
-53.1
cm/s


SAMSUNG
HERA W10
AoV
5

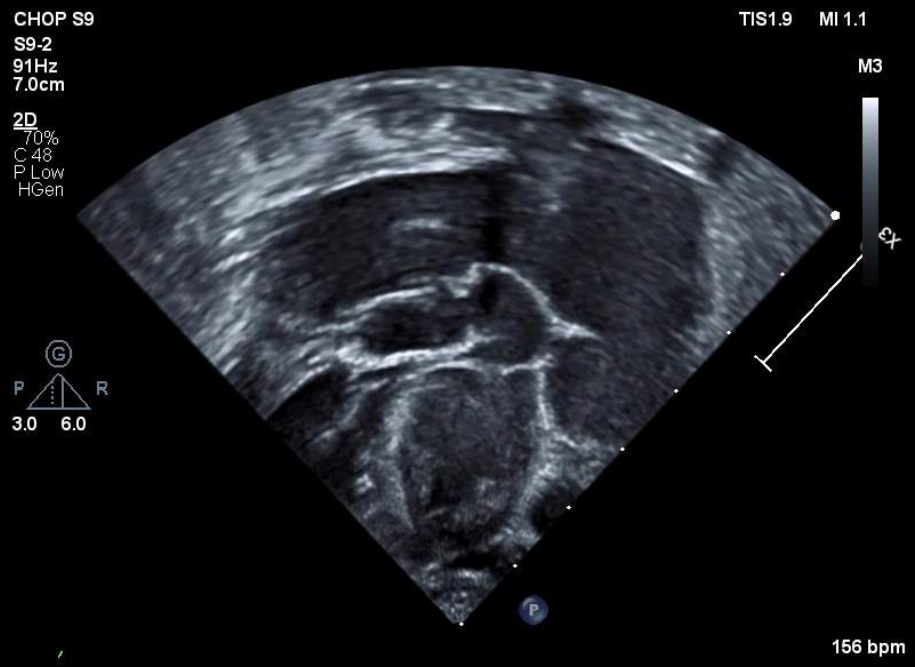

29W6 F/U
VERTEX
Ao_Arch
SAMSUNG
HERAW10
70.7
-70.7
cm/s

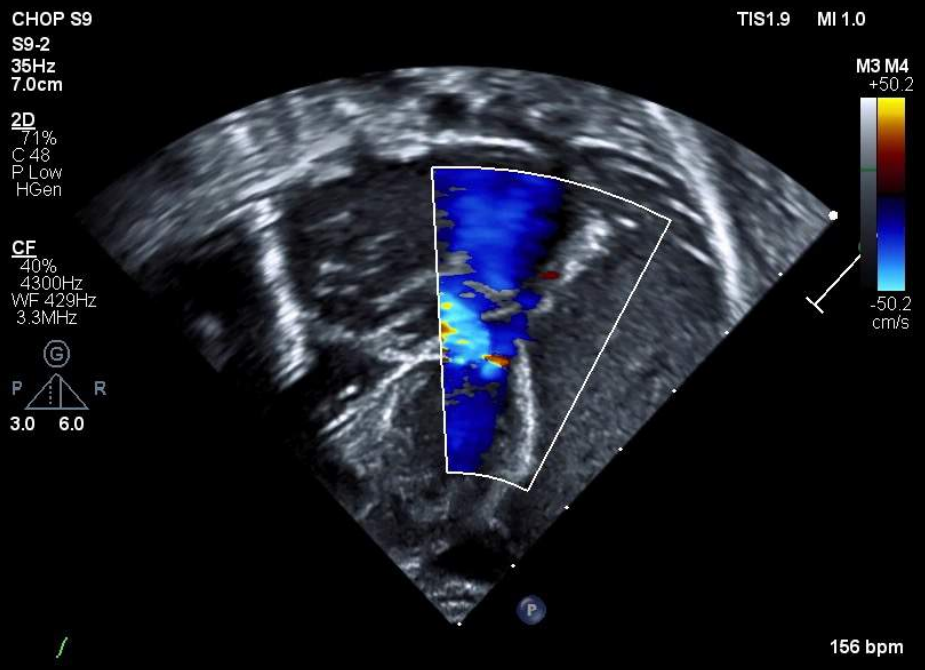

[2D]
Frq Gen.
GN 47
DR 48
FA 3
P 90
29W6 F/U
VERTEX
Ao_Arch
N6 F/U
VERTEX
Ao_Arch
70.7
-70.7
cm/s
GN 50
7.40kHz
F 2
P 90
3.0mm
0°
@6.4cm
1.5
1
0.5
m/s
-0.5

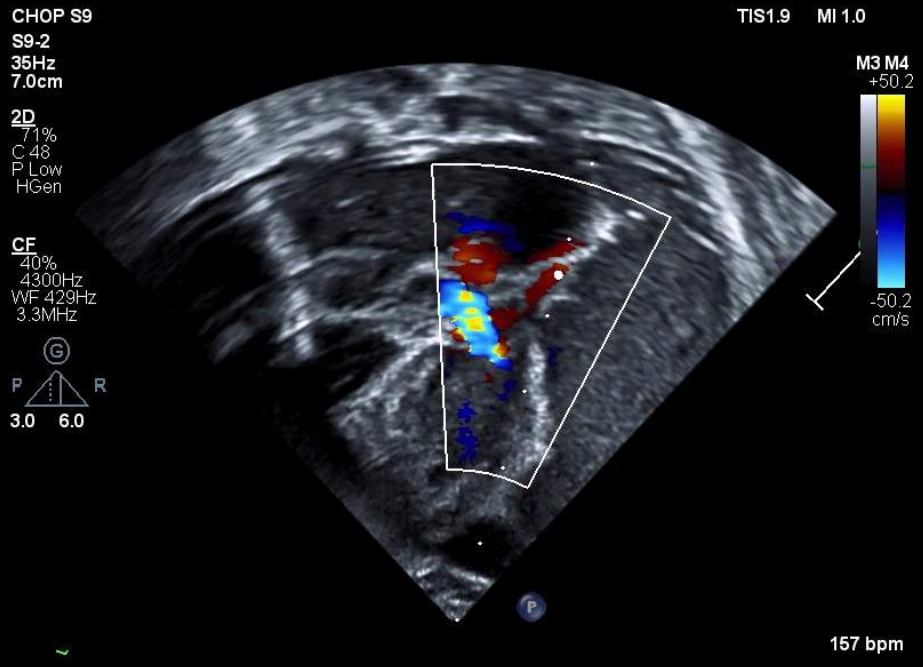


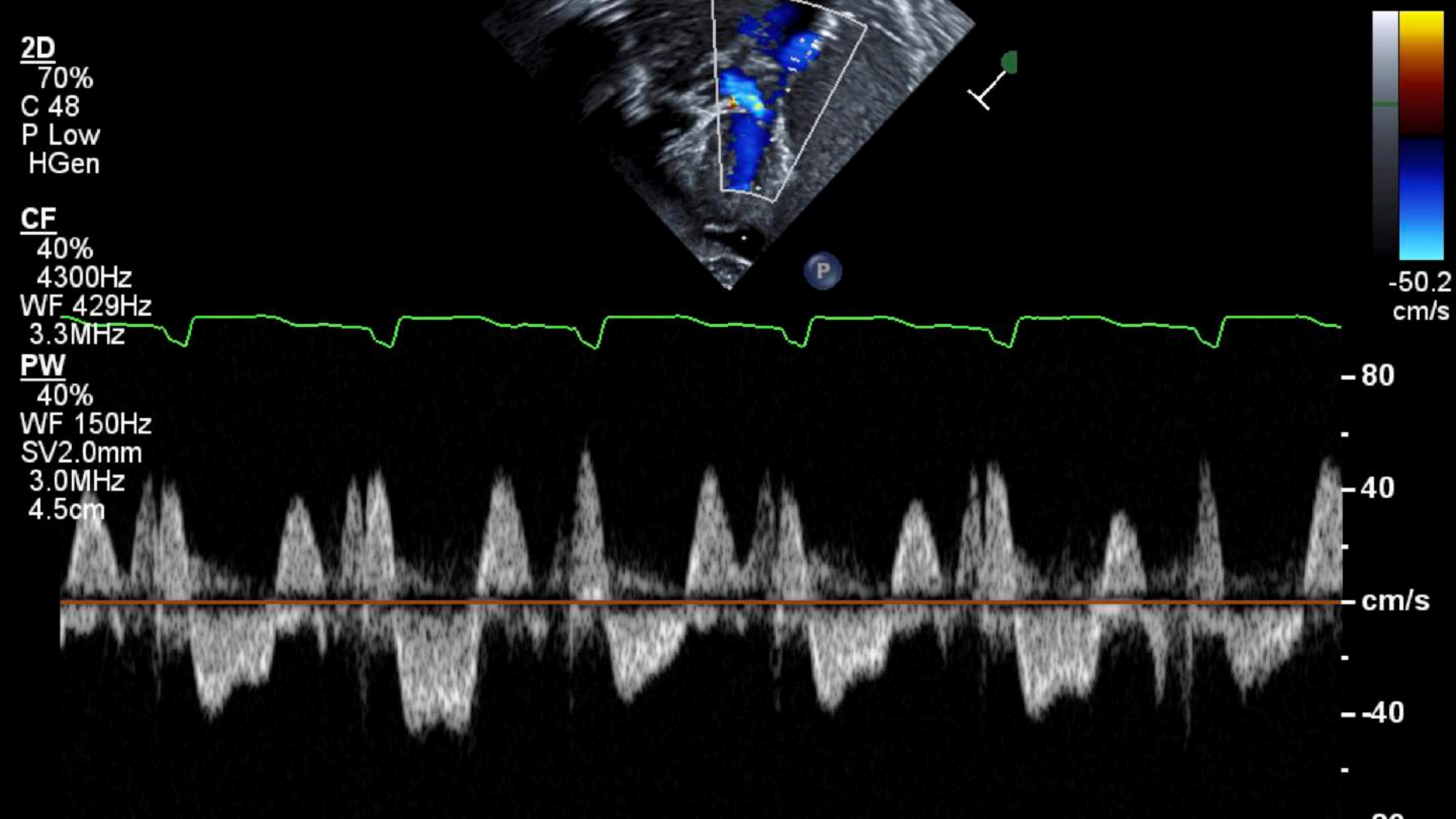

SAMSUNG
HERAW10

Postnatal ECHO



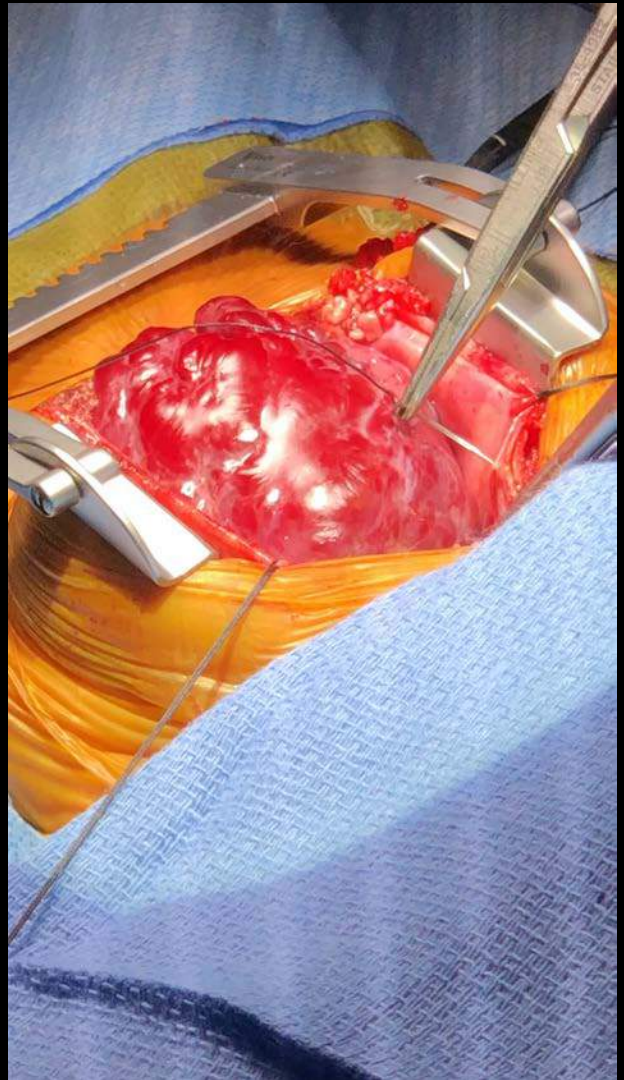

CHOP S9
S9-2
35Hz
7.0cm
2D
71%
C 48
P Low
HGen
CF
40%
4300Hz
WF 429Hz
3.3MHz
G
P
R
3.0
6.0
TIS1.9
MI 1.0
M3 M4
+50.2
-50.2
cm/s
156 bpm

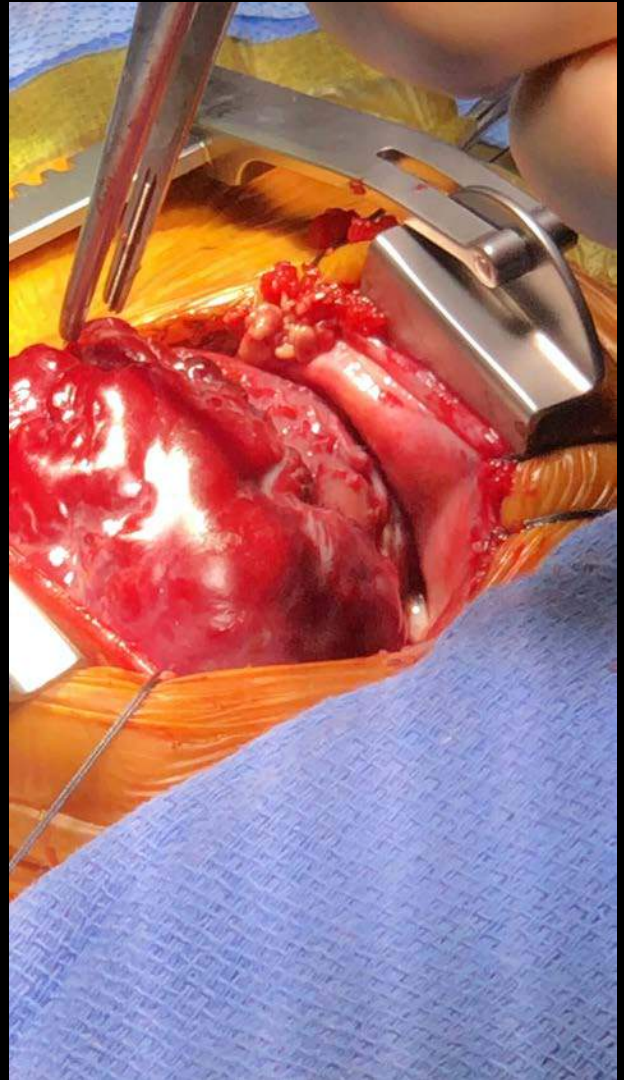

CHOP S9
S9-2
35Hz
7.0cm
2D
71%
C 48
P Low
HGen
CF
40%
4300Hz
WF 429Hz
3.3MHz
G
P
R
3.0
6.0
TIS1.9
MI 1.0
M3 M4
+50.2
-50.2
cm/s
157 bpm


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


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
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.





Gender: Male

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

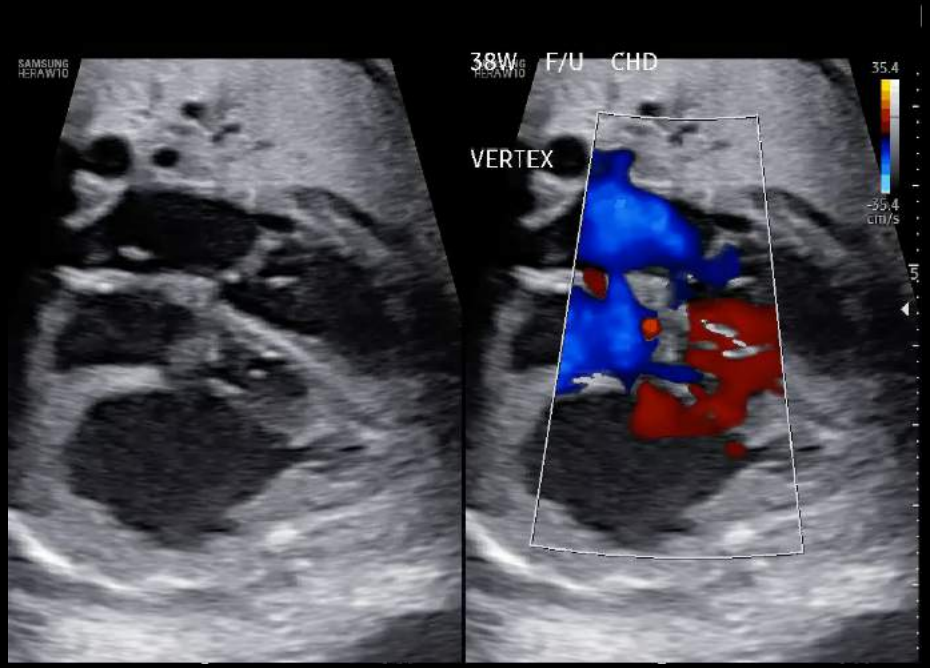
Sample(s) Analyzed:
1: Genomic DNA isolated from blood
2: Genomic DNA isolated from Left foot frozen tissue sample
3: Genomic DNA isolated from Left foot FFPE sample

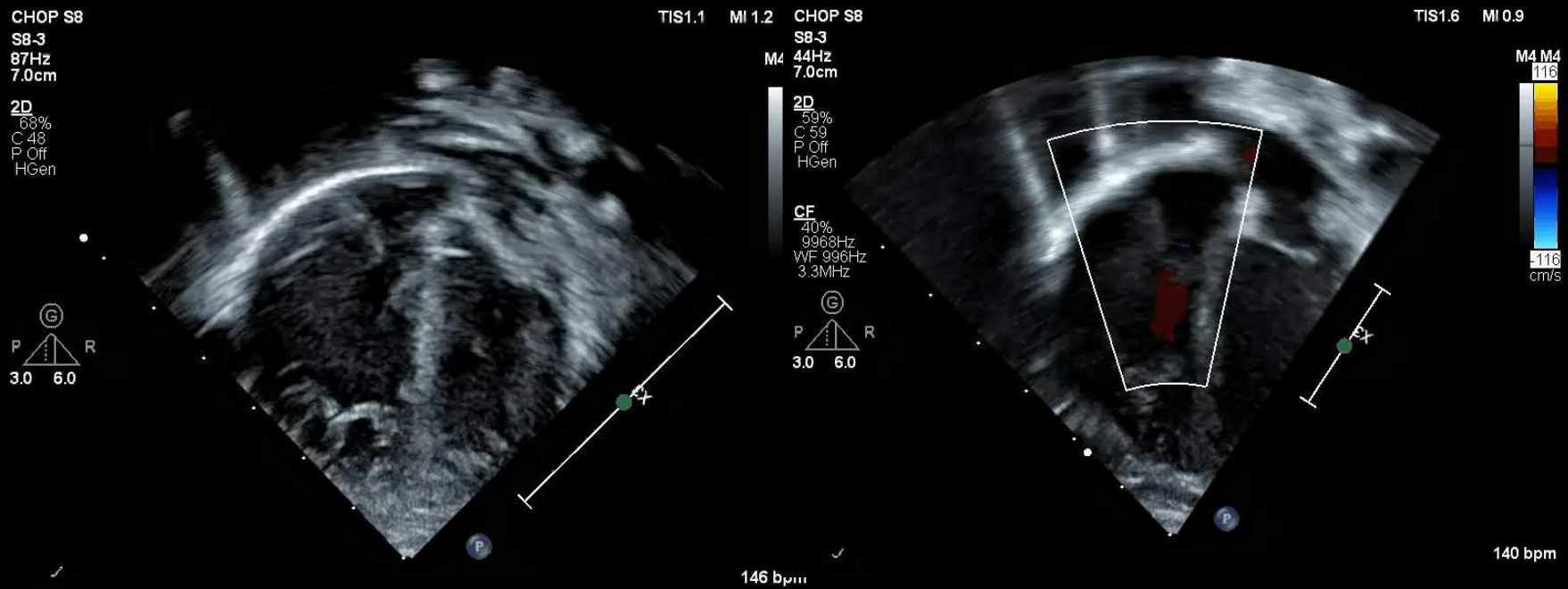
Result Summary: POSITIVE Two Somatic Pathogenic Variants in *TEK*

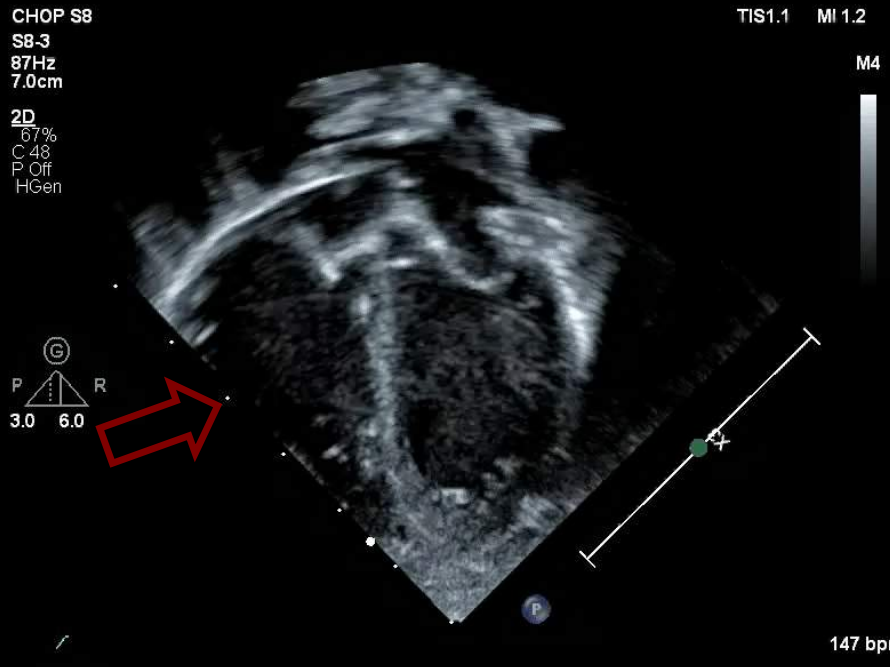
Identification of Variant(s):

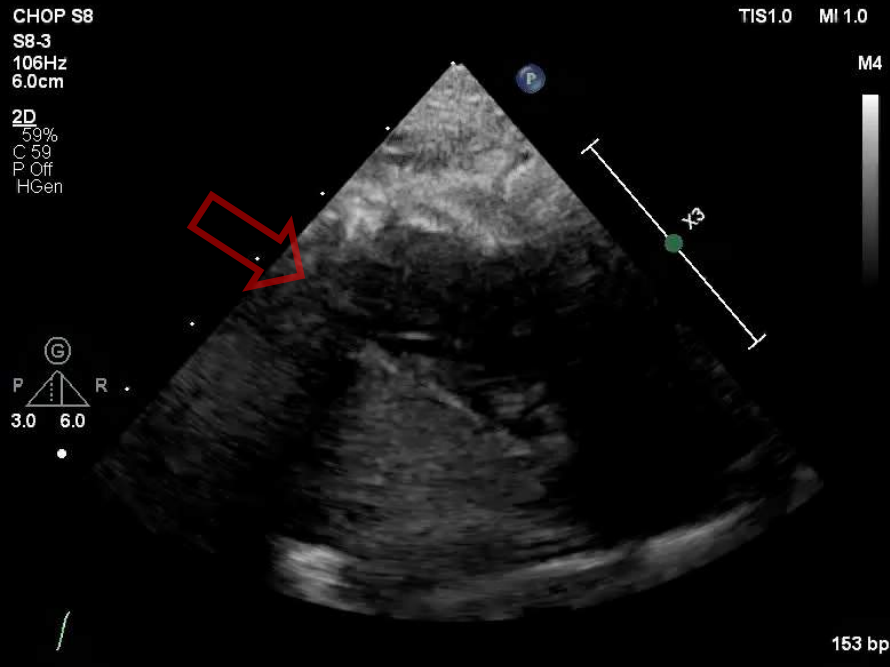
Sample	Gene	Nucleotide Position	Amino Acid Position	Variant Frequency	Classification
2	*TEK*	c.2690A>T	p.Tyr897Phe	10.3 - 12.5%	Pathogenic
2	*TEK*	c.2753G>A	p.Arg918His	9.9 - 13.7%	Pathogenic

CASE 2: Fetus @ 38 weeeks




CHOP S8
S8-3
87Hz
7.0cm
2D
68%
C 48
P Off
HGen
TIS1.1 MI 1.2
M4
P R
3.0 6.0
146 bpm
CHOP S8
S8-3
44Hz
7.0cm
2D
59%
C 59
P Off
HGen
CF
40%
9968Hz
WF 996Hz
3.3MHz
P R
3.0 6.0
TIS1.6 MI 0.9
M4 M4
116
-116
cm/s
140 bpm


CHOP S8
S8-3
87Hz
7.0cm
2D
67%
C 48
P Off
HGen
TIS1.1 MI 1.2
M4
P R
3.0 6.0
147 bpm

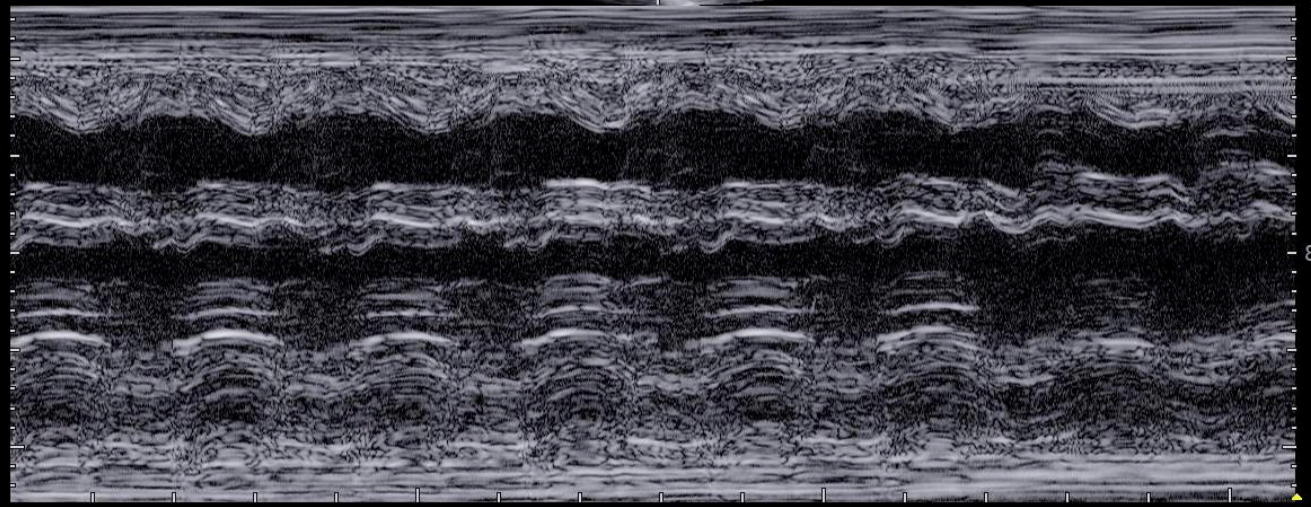

CHOP S8
S8-3
106Hz
6.0cm
2D
59%
C 59
P Off
HGen
TIS1.0 MI 1.0
M4
P R
3.0 6.0
153 bpm

CASE 3


SAMSUNG
HERA W10
53.1
-53.1
cm/s


SAMSUNG
HERA W10
10

[2D]
Frq Gen.
GN 52
DR 48
FA 3
P 90
[M]
GN 50
DR 48
P 90



CASE 4: Presented @ 26 weeks


26W2D
VERTEX


26W2D
VERTEX

Back @ 29 weeks…


CHOP Cardiac C9 New
C9-2
55Hz
S1
2D
75%
Dyn R 48
P Med
HGen
GA 30w1d
Vertex
TIB0.5 MI 0.9
M2
X2
12cm
*** bpm
P
R
2.5 5.0


CHOP Cardiac C9 New
C9-2
60Hz
S1
Z 1.1
2D
73%
Dyn R 48
P Med
HGen
GA 30w1d
Vertex
TIB0.6 MI 0.9
M2
X2
12cm
*** bpm
P
R
2.5 5.0


CHOP Cardiac C9 New
C9-2
83Hz
S1
Z 1.2
2D
77%
Dyn R 48
P Med
HGen
TIB0.9
MI 0.9
M2
X2
*** bpm
G
P
R
2.5
5.0


CHOP Cardiac C9 New
C9-2
95Hz
S1
Z 1.2
2D
73%
Dyn R 48
P Med
HGen
TIB0.9
MI 0.9
M2
X2
*** bpm
P
R
2.5
5.0


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
TIB1.0
MI 0.8
M2 M2
+28.9
-28.9
cm/s
X2
*** bpm
P
R
2.5
5.0

Back again @ 34 weeks


CHOP Cardiac C9 New
C9-2
67Hz
S1
Z 1.1
2D
71%
Dyn R 48
P Med
HGen
GA 34w1d
TIB0.6
MI 0.9
M2
G
P
R
2.5
5.0
X2
10cm
*** bpm


CHOP Cardiac C9 New
C9-2
63Hz
S1
2D
74%
Dyn R 48
P Med
HGen
GA 34w1d
TIB0.6 MI 0.9
M2
X2
G
P
R
2.5 5.0
12cm
*** bpm


CHOP Cardiac C9 New
C9-2
76Hz
S1
2D
71%
Dyn R 48
P Med
HGen
TIB0.7
MI 0.9
M2
X2
10cm
*** bpm
P
R
2.5
5.0