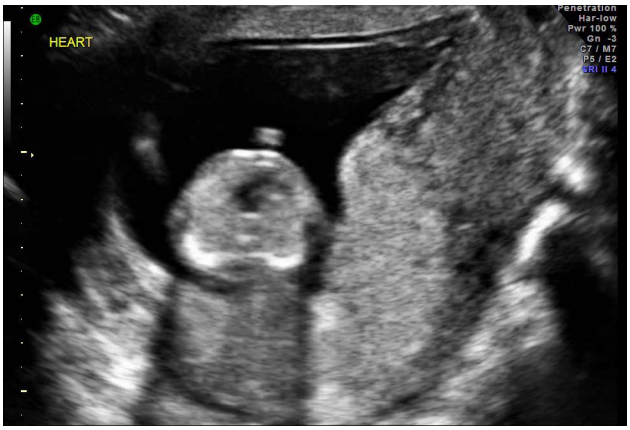
LAURIE REINHARDT
CASE STUDY

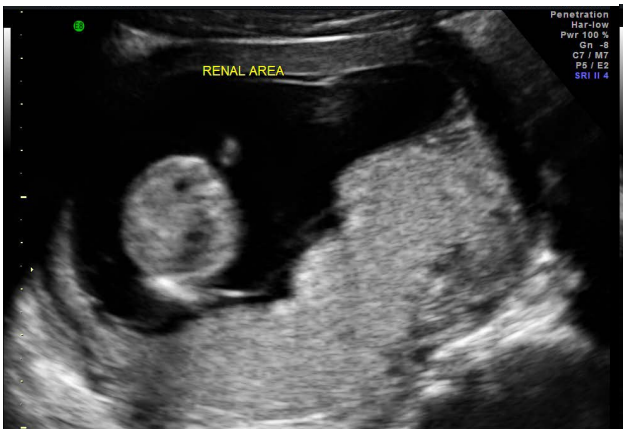
Thirty-seven year old female presented to the sonography department seventeen weeks and two days gestation. Previous amniocentesis revealed female karyotype with Trisomy 21. A karyotype provides a physician with information pertaining to the number of chromosomes, the structure of chromosomes, and the sex of the baby. By obtaining this information the fetus can be accurately diagnosed with chromosomal disorders, such as Trisomy 13, Trisomy 18, and Trisomy 21. Also, patient's lab work identified the fetus with Rh positive.

The patient's sonogram showed the fetus with a possible heart defect with an echogenic foci in left ventricle. Within the abdomen the fetus had echogenic bowel and prominent renal pelves. The images revealed an abnormal face profile and slightly increased nuchal fold. When scanning the fetus's hand, clinodactyly (bending of the fifth digit) was visualized.

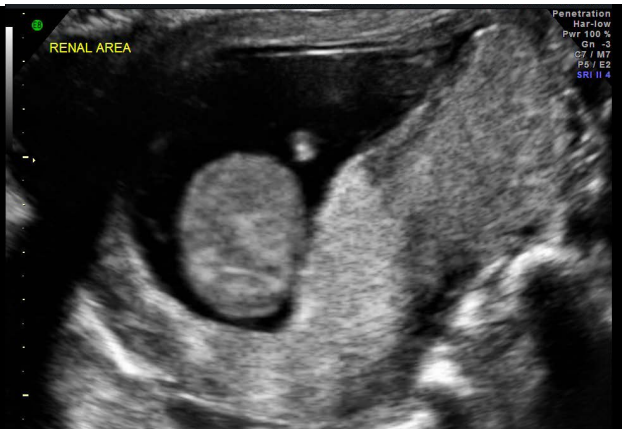
The following images illustrate several soft makers for Trisomy 21.



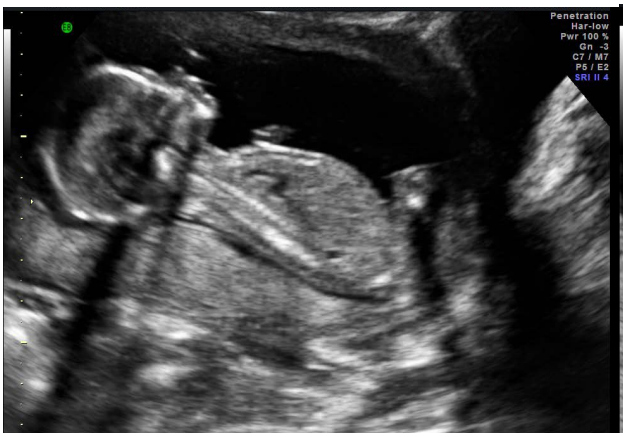
[Image 1: Echogenic Intracranial Focus]

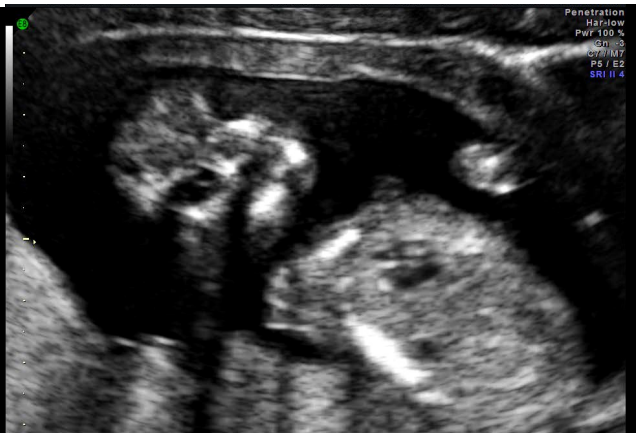


[Image 2: Pyelectasis]

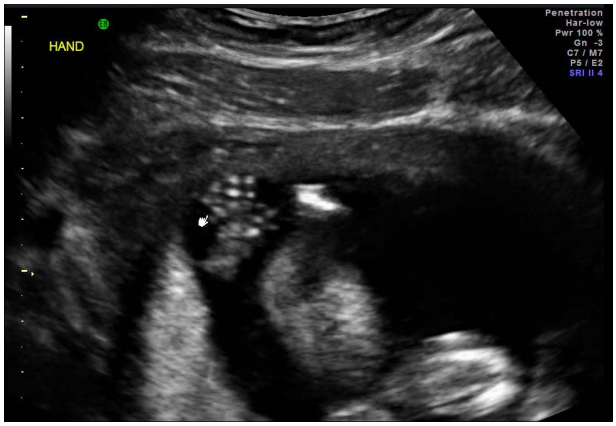


[Image 3: Hyperechoic Bowel]





[Image 4 & 5: Hypoplastic/absent nasal bone]



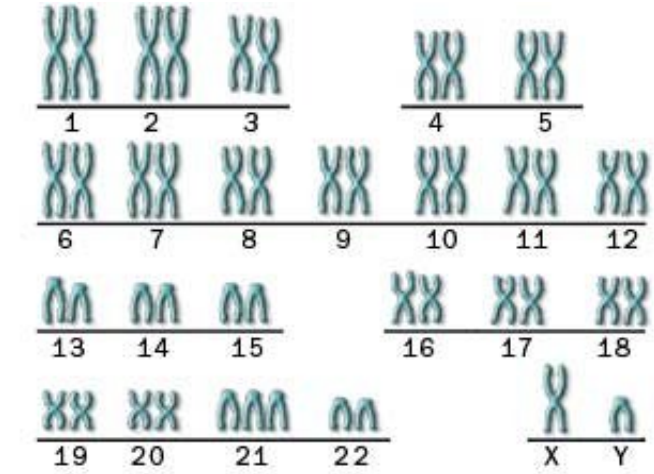
[Image 6: Clinodactyly]

After reviewing the patient's ultrasound and lab results, the physician diagnosed the fetus with Trisomy 21. The most common chromosomal disorder to result in a live birth is Trisomy 21(Down Syndrome).[1] By utilizing the combined information from the karyotype and the sonographic images the physician was able to provide the patient with the knowledge and awareness of the chromosomal disorder that her fetus is diagnosed with. Following the ultrasound the patient and her husband chose to terminate the pregnancy.

SONOGRAPHIC MARKERS FOR TRISOMY 21
Hypoplastic/absent nasal bone
Nuchal fold >5mm
Echogenic intracranial focus
Heart defect
Wide iliac angle
Short femurs
Short humerus
Echogenic/Hyperechoic bowel
Clinodactyly
Duodenal atresia
Pyelectasis (renal pelvis >4mm)
Mild ventriculomegaly

(Rumack[1])

KARYOTYPE with TRISOMY 21



[2]

References

1. Rumack, Carol. Diagostic Ultrasound. 4th ed. Vol.2. Philadelphia: Mosby, 2011:1119.
2. The Genetic Basis of Down Syndrome: 1998-2012 Mayo Foundation for Medical Education and Research. http://www.mayoclinic.com/health/medical/IM02633.