

# Genetic Sonography

## The Ultrasound Detection of Trisomy 21 and Other Chromosomal Abnormalities

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e-mail: fetalecho@fetalecho.com, 2004, CD-ROM, \$450.00

This new multimedia-based program summarizes how sonography can be used to detect genetic disorders of the fetus. Gregory R. DeVore, MD, has developed a set of 4 CD-ROM disks and presents some of the scientific rationale, sonographic methods, and technical pitfalls related to common fetal sonographic markers of aneuploidy.

The main software application includes a myriad of excellent features, such as professional-quality graphics, audio commentary, anatomic labels, a user-friendly interface, navigation bar, help section, and more than 350 video clips. As a minor enhancement, it would have been nice for the clips to automatically play when a video window is first opened. Computer requirements are a Microsoft Windows 98, Me, 2000, or XP operating system, 256 megabytes or more of RAM, and a Pentium 3 or higher processor. A telephone number, fax number, and e-mail address are also provided for technical support. The program offers up to 25 continuing medical education category I credits for this educational activity and fulfills the requirements for continuing medical education and ultrasound.

Software installation is automated by a straightforward procedure. Accessory programs, such as Apple QuickTime 5.0 or higher and Adobe Acrobat Reader are packaged on the installation disk. Although the software is designed to run off a CD-ROM, instructions are provided for transfer of 2.5 gigabytes of data directly to a hard disk. This provides a convenient way to run the entire program from a hard drive. The program includes abstracts and full-text articles (Adobe PDF format) from the medical literature, in addition to a free link to an electronic textbook by the Fetal Medicine Foundation.

Educational content is divided into 2 main sections. The first section, "First-Trimester Nuchal Translucency Screening," covers several tutorials about fetal anatomy, nuchal translucency measurements, 3- and 4-dimensional sonography, fetal nasal bone evaluation, Doppler sonography of the ductus venosus, and maternal serum biochemical markers. Emphasis is placed on early sonographic assessment of the fetal heart. Information is also provided about certification programs for first-trimester screening of genetic abnormalities with their respective Internet links.

A second main section, "Second-Trimester Genetic Sonogram," consists of several tutorials. The Volume 2 tutorial explains how to optimize gray scale and color Doppler settings from various sonography systems, reviews the medical literature regarding sonographic

markers for fetal aneuploidy, provides sonographic examples, and includes a 6-minute sonographic examination of a 17-week fetus. The Volume 3 tutorial reviews 10 unknown cases using motion video clips. Immediate feedback and key teaching points are provided for the user. Volume 4 presents a comprehensive collection of fetuses with abnormalities, including trisomy 13 (n = 3 fetuses), trisomy 18 (n = 16 fetuses), trisomy 21 (n = 36 fetuses), and Turner syndrome (n = 5 fetuses), with their sonographic findings. This tutorial presents more than 1000 randomly selected sonographic images from fetuses with aneuploidy. Video clips can be reviewed, with either slow motion or frame-by-frame controls, and the user tries to identify each abnormality. Immediate feedback is provided after the diagnosis is selected. Another tutorial consists of an image library of abnormal sonographic findings associated with fetal aneuploidy. These abnormalities are linked to a variety of full-text articles or other related Internet links. A final section in Volume 4 provides a practical demonstration and explanation of the statistical approach used to assign genetic risk on the basis of fetal sonographic findings. Concepts such as sensitivity, specificity, likelihood ratios, logistic regression, and prior/posterior probabilities are defined. Dr DeVore has included a "Risk Calculator" to determine a patient's individual risk for chromosomal abnormalities such as trisomy 18 and trisomy 21 and an adjusted risk based on sonographic results. The user is given the option to use risk data from either the author or from other sources as well.

Overall, the program nicely summarizes key literature, explains sonographic technique and pitfalls, presents normal and abnormal cases, and lists Web-based resources for certification programs. Excellent opportunities are provided for improving pattern recognition skills related to the sonographic detection of fetal anomalies. Finally, Dr DeVore provides a practical tool, with simplified explanations of complex statistical concepts, that is very helpful for calculating genetic risk. This is a comprehensive multimedia learning curriculum that is highly recommended for health care practitioners who wish to update their understanding about how sonography can be used to evaluate fetuses at risk for common genetic disorders.

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