

Integrated Screen Cut-off Change for Down syndrome

Effective Date: April 18, 2016

The full and serum integrated screen cut-offs for trisomy 21 will be changed from the current 1 in 110 risk to 1 in 270 - this is in keeping with some of the current national laboratory cut-offs. Based on our full integrated screen data from the past 2-1/2 years, this change will increase the positive rate from its current 1.6%, to 3.4%. Our detection rate for trisomy 21 is then predicted to be approximately 95%. This decision has been made by both OB/GYN and Beaumont Lab leadership.

When Beaumont adopted the full integrated screen (ref) for routine clinical use, we took advantage of the high specificity to reduce our number of false positive results, while still maintaining a reasonably high detection rate. Since inception, our cut off for a positive test has been a trisomy 21 risk of 1 in 110 or higher, resulting in a 1–2 percent positive rate while maintaining an estimated detection rate of slightly less than 90%. Additional benefits of the full integrated screen have included:

1. A first trimester ultrasound exam to evaluate both for dating and potential early detection of severe anomalies
2. Unexpected inclusion of other chromosomal abnormalities captured by the abnormal full integrated screen

However, the recent availability of cell free DNA testing has dramatically changed how patients approach genetic screening and testing. Although an amniocentesis is still indicated to follow-up an abnormal full integrated screen, many patients are choosing to evaluate an abnormal screen with DNA testing, preferring the safety of this approach despite the higher abnormal karyotype detection rate afforded by amniocentesis. However it should be noted that cell-free DNA testing is still regarded by ACOG as a screen and not a diagnostic test.

Reference: N Engl J Med 2005;353:2001-11

If you have questions, please contact Client Services (1-800-551-0488, option 5).

Laboratory Test Directory: <http://beaumontlaboratory.com/test-lab-directory>.

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Beaumont Laboratory

Customer Service

1-800-551-0488