# What is BabySEQ?

BabySEQ is a Non-Invasive Prenatal Testing (NIPT) method, a new-generation screening that detects not only chromosomal disorders such as Down syndrome, Trisomy 13, and Trisomy 18, but also microdeletion syndromes in the fetus, without the need for any invasive procedure that may involve risk. Currently, NIPT-based tests like BabySEQ are known to be the most accurate tests for screening these conditions. BabySEQ, which is performed in our center, has become an increasingly preferred screening method in recent years due to its non-invasive nature and ease of application. BabySEQ includes four different panels, depending on your preference.

| | BabySEQ BASIC PANEL | BabySEQ EXTENDED PANEL | BabySEQ RARE* | BabySEQ XL* |
|---|---|---|---|---|
| Common Aneuploidies (T21, T18, T13, X/Y) | ✓ | ✓ | ✓ | ✓ |
| All Other Aneuploidies | ✓ | ✓ | ✓ | ✓ |
| Deletion and Duplication Syndromes | | ✓ | ✓ | ✓ |
| Cystic Fibrosis + SMA + Fragile X Carrier Screening | | | ✓ | ✓ |
| Carrier Screening for 700 Diseases | | | | ✓ |

For these tests, it is recommended to collect one tube of blood from the father (purple-top EDTA tube) and one tube of blood from the mother using the BabySEQ blood collection tube provided by Intergen.

**What is BabySEQ RARE?**

In addition to the extended BabySEQ panel, the BabySEQ RARE package also screens for SMA, Cystic Fibrosis, and Fragile X Syndrome.
Scan the QR code to view the BabySEQ RARE Information Brochure.

In the BabySEQ Rare package, carrier testing is also performed for the father.

**What is BabySEQ XL?**

The BabySEQ XL package includes the Extended Panel, which examines the fetus for all chromosomal anomalies.
In addition, it provides screening of the prospective parents for 700 genetic conditions.
Scan the QR code to view the BabySEQ XL Information Brochure.

In the BabySEQ XL package, carrier testing is also performed for the father.

**For detailed information about the BabySEQ Rare and XL packages, please consult your physician.**

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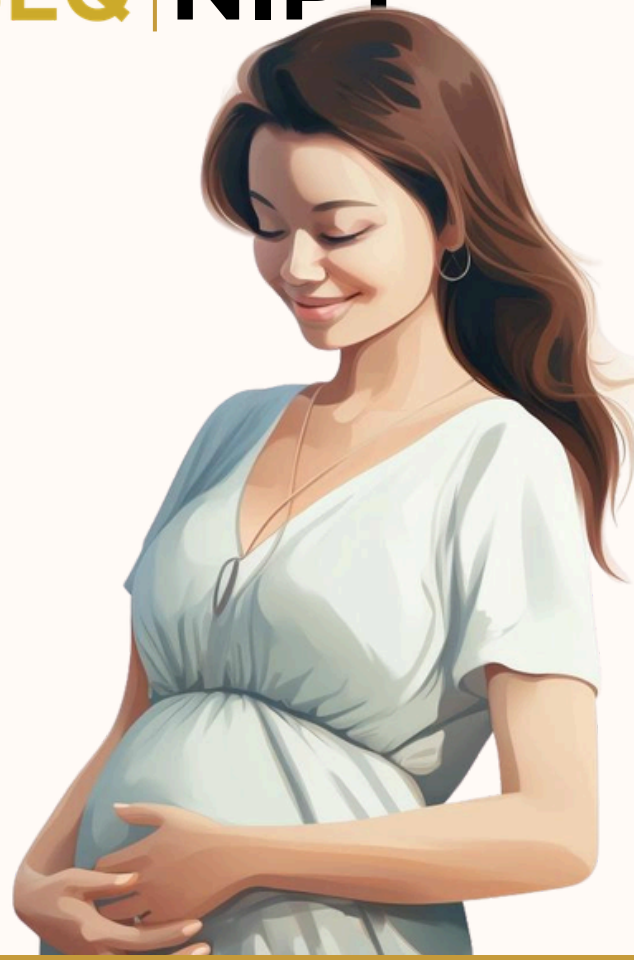
# babySEQ | NIPT



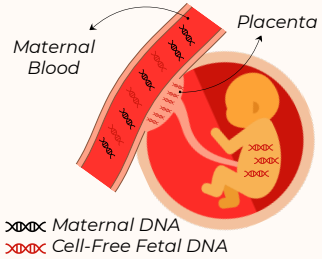
# What is Being Tested?

The most common autosomal and gonosomal aneuploidies are shown below. **The Basic Panel** screens for all chromosomal aneuploidies.

| Aneuploidy | | |
|---|---|---|
| Trisomy 21 (Down Syndrome)<br>Trisomy 18 (Edwards Syndrome)<br>Trisomy 13 (Patau Syndrome)<br>45, X (Turner Syndrome)<br>47, XXY (Klinefelter Syndrome)<br>47, XXX | Trisomy 7<br>Trisomy 8<br>Trisomy 9<br>Trisomy 10<br>Trisomy 11<br>Trisomy 12<br>Trisomy 14 | Trisomy 15<br>Trisomy 16<br>Trisomy 17<br>Trisomy 19<br>Trisomy 20<br>Trisomy 22<br>Other Aneuploidies |

**In the Extended Panel,** in addition to all chromosomal aneuploidies, deletion/duplication syndromes are also screened.
For screening purposes, deletion/duplication syndromes ≥ 7 Mb in size are evaluated.
The list of deletion/duplication syndromes screened is shown on the right.

**Note: Apart from the changes defined in the table, if any alteration greater than 7 Mb is detected, it will be indicated in the report.**




During pregnancy, fragments of fetal DNA, predominantly originating from the placenta, pass into the mother's circulation. After the 10th week of pregnancy, with the BabySEQ NIPT method, we can analyze this fetal DNA present in the maternal bloodstream.

## Deletion/Duplication Syndromes

1p32-p31 Deletion Syndrome
1q41-q42 Deletion Syndrome
1q43-q44 Deletion Syndrome
2p12-p11.2 Deletion Syndrome
2p15-p16.1 Deletion Syndrome
2q13 Deletion Syndrome
2q13 Duplication Syndrome
2q31.1 Microdeletion Syndrome
2q31.1 Duplication Syndrome
2q35 Duplication Syndrome
3p25.3 Deletion Syndrome
3pter-p25 Deletion Syndrome
3q13.31 Deletion Syndrome
Dandy-Walker Syndrome (3q Deletion/Duplication)
3q26 Microduplication Syndrome
3q29 Deletion Syndrome
4q21 Deletion Syndrome
Axenfeld-Rieger Syndrome, Type L (4q25)
5p13 Duplication Syndrome
5q12 Deletion Syndrome
5q14.3 Deletion (Proximal) Syndrome
Sotos Syndrome (5q35 Deletion)
6p22 Microdeletion Syndrome
6q11-q14 Deletion Syndrome
6q24-q25 Deletion Syndrome
Coffin-Siris Syndrome 1 (6q25)
Greig Cephalopolysyndactyly Syndrome (7p14)
7p22.1 Microduplication Syndrome
7q11.23 Deletion (Distal) Syndrome
Williams-Beuren Syndrome (WBS) (7q11.23)
Currarino Syndrome (7q36)
7q36.3 Duplication Syndrome
8p11.2 Deletion Syndrome
8p23.1 Deletion Syndrome
8q12 Microduplication Syndrome
Nablus Mask-Like Facial Syndrome (NMLFS) (8q22.1)
**Trichorhinophalangeal Syndrome, Type 2 (TRPS2) (8q23.3-q24.11)**
**9p Deletion Syndrome**
**9p13 Microdeletion Syndrome**
**9p24.3 Deletion Syndrome**
**9q33.33-q34.11 Microdeletion Syndrome**
**Kleefstra Syndrome 1 (KLEFS1) (9q34.3)**
**10p11.21-p12.31 Microdeletion Syndrome**
**DiGeorge Syndrome – Velocardiofacial Syndrome Complex 2 (DSG2) (10p14)**
**10q22.3-q23.2 Deletion Syndrome**
**Split Hand/Foot Malformation 3 (SHFM3) (10q24)**
**10q26 Deletion Syndrome**
**Potocki-Shaffer Syndrome (11p12-p11.2)**
**WAGR Syndrome (11p13)**
**WAGRO Syndrome (11p13-p14)**
**11q13.2-q13.4 Deletion Syndrome**
**11q22.2-q22.3 Microdeletion Syndrome**
**11q23 Deletion Syndrome**
**Distal Trisomy 11q Syndrome**
**12p12.1 Microdeletion Syndrome**
**12q14 Microdeletion Syndrome**
**12q15-q21.1 Microdeletion Syndrome**
**13q14 Deletion Syndrome**
**14q11-q22 Deletion Syndrome**
**Frias Syndrome (14q22-q23)**
**14q24.1-q24.3 Microdeletion Syndrome**
**Prader–Willi Syndrome (15q11-q13)**
**15q13.3 Deletion Syndrome (BP4/BP5)**
**15q14 Microdeletion Syndrome**
**15q25.2 Deletion (Proximal) Syndrome**
**15q26-qter Deletion Syndrome**
**16p11.2-p12.2 Microdeletion Syndrome**
**16p12.2 Deletion (Proximal) Syndrome**
**16p13.11 Duplication Syndrome**
**16p13.11 Deletion Syndrome**
**Rubinstein-Taybi Syndrome (16p13.3)**
**Alpha Thalassemia–Intellectual Disability Syndrome linked to Chromosome 16 (ATR-16 Syndrome) (16p13.3)**
16q22 Deletion Syndrome
Smith-Magenis Syndrome (17p11.2)
Yuan-Harel-Lupski Syndrome (YUHAL) (17p12-p11.2)
17p12 Deletion Syndrome
17p12 Duplication Syndrome
17p13.1 Deletion Syndrome
Miller-Dieker Lissencephaly Syndrome (MDLS) (17p13.3) (Deletion)
Miller-Dieker Lissencephaly Syndrome (MDLS) (17p13.3) (Duplication)
17p13.3 Telomeric Duplication Syndrome
17q12 Deletion Syndrome
17q21.31 Deletion Syndrome
17q23.1-q23.2 Deletion Syndrome
Tetrasomy 18p Syndrome
18p Deletion Syndrome
18q Deletion Syndrome
19p13 Duplication Syndrome
19q13.11 Microdeletion Syndrome
20p13 Microdeletion Syndrome
21q22.11-q22.12 Microdeletion Syndrome
DiGeorge Syndrome – Velocardiofacial Syndrome (22q11.2) (Proximal)
22q11.2 Deletion Syndrome (Distal)
22q13 Deletion Syndrome
22q13 Duplication Syndrome
Xp11.22 Duplication Syndrome
Xp11.22-p11.23 Duplication Syndrome
Xp11.23 Microdeletion Syndrome
Xp11.3 Deletion Syndrome
Xp21 Microdeletion Syndrome
Xp21.2 Microduplication Syndrome
Xp22.31 Microdeletion Syndrome
Xq21 Microdeletion Syndrome
Xq22.3 Telomeric Deletion Syndrome
Xq27.3-q28 Duplication Syndrome
Xq28 Deletion Syndrome

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