

1 What is NACE®?

- NACE® is a **non-invasive prenatal screening test** that analyzes the most frequent chromosomal abnormalities **without compromising the pregnancy**, from week 10.
- A simple fetal blood draw from the mother allows free DNA circulating in the maternal bloodstream to be detected via next generation sequencing (NGS) technology and proprietary bioinformatic analysis.



Test carried out in Spain

2 What does NACE® detect?

- NACE® detects **abnormalities in chromosomes 21, 18, 13** and the most common anomalies in the sexual chromosomes (X and Y).

NACE® 24 analyzes **all chromosomes**.

NACE® 24 Extended analyzes **all 24 chromosomes** and identifies **microdeletions** associated with **6 major genetic syndromes**.

	NACE®	NACE® 24	NACE® 24 Extended
Down syndrome	✓	✓	✓
Edwards syndrome	✓	✓	✓
Patau syndrome	✓	✓	✓
Sexual chromosomes	✓	✓	✓
All remaining chromosomes		✓	✓
Microdeletions			✓
Turnaround time	3 days	4 days	10 days

Sexual chromosomes:

- Turner syndrome (45, X)
- Klinefelter syndrome (XXY)
- XYY syndrome
- X trisomy syndrome

For twin pregnancies, sexual chromosomes are not analyzed.

Microdeletions:

- DiGeorge syndrome
- Angelman syndrome*
- Cri-du-chat syndrome
- p36 deletion syndrome
- Prader-Willi syndrome*
- Wolf-Hirschhorn syndrome

* The microdeletion region is the same for both Angelman and Prader-Willi syndromes (15q11.2). NACE 24* Extended does not distinguish between the two syndromes. An additional confirmation test will be required.

3 Who is NACE® suitable for?

- Several scientific support the use the NACE test for all pregnant women⁽¹⁾.

It is particularly recommended for women with:

- An abnormal result in their first trimester screen
- A previous Down's syndrome pregnancy
- A suspicious ultrasound finding

Gregg, et al. Genet Med. 2016

4 Cases where NACE® can be used

- Single pregnancies
- Twin pregnancies
- IVF
- Egg donation
- Women of all ages
- Women of all ethnicities
- Women of all body mass index
- Consanguinity

1. Gregg AR et al. Genet Med 2016; 18:1056-65.
2. Bianchi et al. N Engl J Med. 2014 27;370(9):799-808.
3. Nicolaides KH. Prenat Diagn 2011; 31:7-15.

* Starting count from the day after the sample is received at Igenomix.

4 Why use a non-invasive prenatal test?

- Non-invasive tests can prevent the need for about 98% of invasive tests in patients at risk for trisomy 21.⁽²⁾
- NACE® reduces the number of miscarriages caused by amniocentesis or corionic villus sampling.



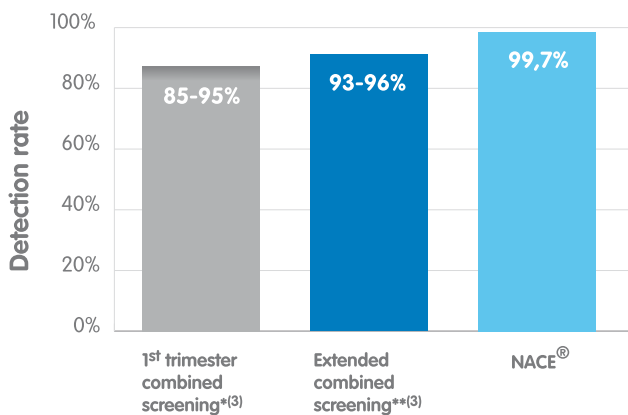
Conventional tests including biochemical screening can lead to:

1 in every 20 women who tests positive for Down syndrome will not be carrying an affected baby.

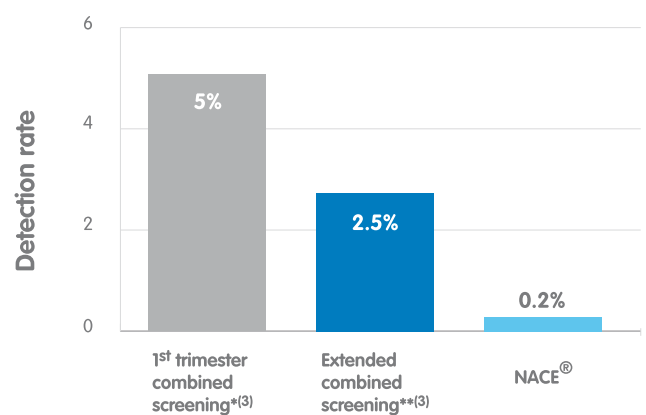
False sense of security

Of every 20 women carrying a baby with Down syndrome, 3 will test negative by biochemical screening.

Down syndrome detection rate



False positive rate according to the type of screening



* Includes maternal age, nuchal translucency measurement, and the detection of the PAPP-A and free β-hCG biochemical markers. Risk assessment can vary depending on diverse factors.

** Includes other ultrasound markers: nasal bone absence, assessment of the ductus venosus, and tricuspid blood flow.

Coverage of the NACE® test for single pregnancies ordered by importance

- According to data from the 2012 European Registry for Prenatal Diagnosis⁽²⁾, abnormalities in chromosomes 21, 18, and 13 represent 71% of all chromosomal abnormalities detected.

