IONA® Nx
NIPT Workflow

Yourgene Health

Innovative NIPT solution for *your* clinical laboratory

The scalable and flexible solution to fit your laboratory throughput:

- Optimal Clinical Menu
- High performance CE-IVD screening
- Fetal Fraction Enrichment

End-to-end NIPT solution
for *your* clinical laboratory

The IONA® Nx Workflow offers a simplified, flexible and scalable solution to suit all laboratories offering NIPT.

A fully validated, CE-marked test that applies whole-genome shotgun sequencing using Next Generation Sequencing technology (NGS) on the Illumina NextSeq™ 550Dx instrument.

Why choose IONA® Nx?

- Innovative: **first NIPT workflow to enrich fetal fraction by up to two-fold** thanks to automated gel size-selection on the Yourgene® QS250 instrument.
- Flexible and scalable workflows for up to 96 patient samples per flow cell
- World-class global technical services team
- Full traceability and data management from start to finish with the Atlas Workflow Manager
- Fast turnaround time from as little as 2 days thanks to rapid handling and overnight processing
- Access to our service labs for back-up and additional capacity
- Cost-effective: minimal capital footprint and high sequencing efficiency

2% FETAL FRACTION

>99% SPECIFICITY

2 DAY WORKFLOW*

A comprehensive menu: whole-genome screening

CORE

- Trisomy 21 (Down's syndrome)
- Trisomy 18 (Edwards' syndrome)
- Trisomy 13 (Patau's syndrome)

OPTIONAL

- Fetal sex determination
- Sex Chromosome Aneuploidies (SCAs)
  - 45,X (Turner syndrome)
  - 47,XXX (Trisomy X)
  - 47,XXY (Klinefelter syndrome)
  - 47,XYY (Jacobs syndrome)
- All additional trisomies
- All monosomies

MICRODELETIONS

IONA® Nx screens for 5 clinically significant microdeletions (≥3 Mb) associated with the following conditions:

- 22q11.2 deletion syndrome – DiGeorge syndrome
- 15q11.2-q13 deletion – Prader-Willi & Angelman syndrome
- 1p36 deletion syndrome
- 5p15- deletion – Cri-du-Chat syndrome
- 4p16.3 deletion – Wolf-Hirschhorn syndrome

*On a fully automated workflow. 3 days for manual workflow.

① Yourgene® SP150

The IONA® Nx NIPT Workflow streamlines the entire process from sample preparation to report generation, enabling greater efficiency. The process begins with spinning the blood down to plasma, following which DNA extraction, amplification and preparation is performed on a single instrument; the Yourgene® SP150. The SP150 utilises unique, fully validated scripts from Yourgene, and is a cost-effective, multi-purpose instrument, designed to automate routine tasks with minimal capital footprint.

Quantification and innovative enrichment of the fetal DNA is then carried out on the Yourgene® QS250 instrument. All platforms are combined for consistent and reliable walkaway automation.

② Yourgene® QS250

The Yourgene® QS250 offers a game-changing NIPT fetal fraction enrichment step with enhanced sensitivity and specificity, especially for low fetal fraction samples.

Laboratories will appreciate the ability to accomplish DNA gel size selection, fragment size analysis and DNA quantification via solution-based fluorescence assay with a single piece of equipment.

Integration of the QS250 with partnering automation in the IONA® Nx NIPT Workflow affords users plug-and-play automation of assay setup and execution which reduces re-draw rate.

IONA® NX AUTOMATED WORKFLOW*

DNA EXTRACTION & LIBRARY CONSTRUCTION

① 

DNA QUANTIFICATION & ENRICHMENT

② 

ATLAS WORKFLOW MANAGER - FULL SAMPLE TRACKING

Quality

The IONA® Nx NIPT Workflow is a CE-marked *in vitro* diagnostic product which meets the requirements laid out in the European In Vitro Diagnostic Medical Devices Directive (98/79/EC).
In addition, the IONA® Software for analysis has been built to stringent quality standards and has been developed with BS EN 62304 compliance.
The IONA® Nx NIPT Workflow received its CE mark in June 2020. This ensures a high-quality robust, reproducible screening test with verification and clinical validation already performed to enable fast implementation with confidence.

Atlas Workflow Manager

The only NIPT solution offering full sample traceability thanks to the Atlas Workflow Manager. This Yourgene Data Management System (DMS) software supports the administrative and technical management of your laboratory samples and data.
The Atlas Workflow Manager tracks your samples and reagents from sample intake to results analysis. It also offers seamless connectivity to your LIMS system for sample/patient information importation and exportation, not only saving time but also reducing potential data entry errors.

*IONA® Nx is also available in a manual workflow

③ Illumina Next Seq 550Dx

The IONA® Nx NIPT Workflow sequencing step is performed using the Illumina NextSeq™ 550Dx instrument

- Upstream IONA® Nx processing means a call can be made from a minimum of 150,000 unique GC-corrected size weighted reads
- Unlike other NIPT providers, our run control is added prior to sequencing, allowing more patients on each flow cell and no waste of sequencing reagents

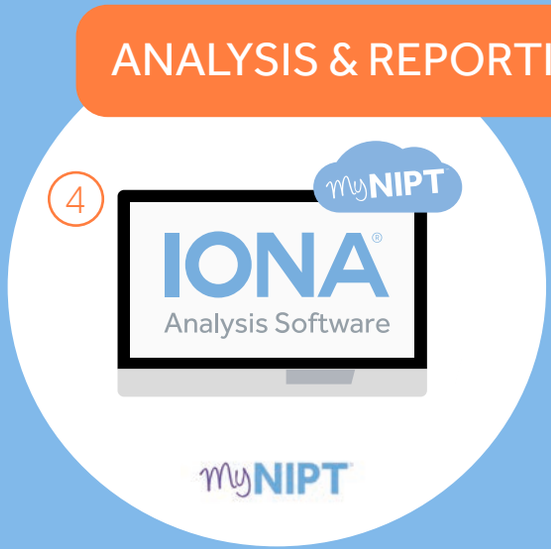
④ IONA® Analysis Software

Yourgene IONA® Analysis Software makes secure, local analysis possible

- Highly efficient, multi-core analysis algorithms are employed which analyse the relative amount of DNA from chromosomes 21, 18 and 13 to calculate a risk score for the presence of a trisomy
- IONA® Software consolidates compounding risk factors including maternal age to produce an adjusted probability of the fetus being affected, without the need for additional bioinformatics interpretation of raw data
- A clinical results report is generated for each sample
- Screening test results can be sent to clinics using the MyNIPT® portal, our safe and secure data exchange portal

SEQUENCING

ANALYSIS & REPORTING



ATLAS WORKFLOW MANAGER - FULL SAMPLE TRACKING

Bespoke reporting software

IONA® Nx results

The relative amounts of chromosomes are used to calculate a test risk score which predicts the presence of an aneuploidy. To personalise the results to each individual patient, this is integrated with a prior risk, such as the age of the mother or the result from the First Trimester Combined Test (FTCT).

IONA® Nx report

The IONA® Nx report gives a clear, easy to interpret, result of high risk or low risk for each trisomy. We recommend that high risk results should be discussed with a healthcare professional in the context of all available clinical findings, including advising for the need for genetic counselling or additional diagnostic testing (e.g. amniocentesis).

SCREENING TEST RESULTS			
TRISOMY	BACKGROUND RISK	The IONA® Nx RISK SCORE	CLINICAL SUMMARY
TRISOMY 21 (Down's Syndrome)	1 : 15	Less than 1 : 10,000 (<0.01%)	LOW RISK
TRISOMY 18 (Edwards' Syndrome)	1 : 31	Less than 1 : 10,000 (<0.01%)	LOW RISK
TRISOMY 13 (Patau's Syndrome)	1 : 100	Less than 1 : 10,000 (<0.01%)	LOW RISK

Estimated Fetal Fraction	5%

Snippet taken from IONA® Nx screening report

Performance you can trust

The clinical performance of the IONA® Nx NIPT Workflow was assessed as part of the development process prior to product launch. Post-launch, we conduct post-market surveillance, which includes a set of activities conducted by manufacturers to collect and evaluate experience gained from medical devices that have been placed on the market.

Global IONA® Test performance observed following Post-Market Surveillance on 76,989 singleton, mono & dichorionic twin pregnancies.

Post-Market Surveillance (PMS) Data

Core Trisomies [a]

Condition	Sensitivity	Specificity	Positive Predictive Value (PPV)	Negative Predictive Value (NPV)
Trisomy 21 (1362 / 76,989)	99.49% (1,355 / 1,362) 95% CI: 98.95-99.79%	99.99% (75,621 / 75,627) 95% CI: 99.98-100%	99.56%	99.99%
Trisomy 18 (430 / 76,989)	97.91% (421 / 430) 95% CI: 96.73-99.35%	99.99% (76,548 / 76,559) 95% CI: 99.97-99.99%	97.45%	99.99%
Trisomy 13 (203 / 76,989)	99.51% (202 / 203) 95% CI: 97.30-99.99%	99.99% (76,781 / 76,786) 95% CI: 99.98-100%	97.58%	>99.99%

[a] Observed performances are based on Post-Market Surveillance of the IONA® Nx workflow in over 76,989 singleton, monochorionic & dichorionic twin pregnancies, from a population of women who are predominantly at a higher risk of having a fetus with Down's syndrome (see prevalence for the population tested). Performances are dependant of laboratories fully reporting discordant results to Yourgene Health as they occur. Please note that the IONA® Nx NIPT Workflow is a Non-Invasive Prenatal Test (NIPT) based on analysis of cell-free fetal DNA (cffDNA) in maternal blood. From data held on file by Yourgene Health. Correct as of 31st Oct 2023.

SCA, AA & Microdeletions [b]

Condition	Sensitivity	Specificity	Positive Predictive Value (PPV)	Negative Predictive Value (NPV)
SCA (157 / 13,924)	98.73% (155 / 157) 95%CI: 95.53-99.85%	99.96% (13,762 / 13,767) 95%CI: 99.92-99.99%	96.88%	99.99%
AA (75 / 12,006)	>99.99% (75 / 75) 95%CI: 95.20-100%	99.96% (11,926 / 11,931) 95%CI: 99.93-99.99%	96.15%	>99.99%
Microdeletions (10 / 747)	>99.99% (10 / 10) 95%CI: 69.15-100%	>99.99% (737 / 737) 95%CI: 99.50-100%	>99.99%	>99.99%

[b] Observed performances are based on Post-Market Surveillance of the IONA® Nx workflow in singleton and monochorionic twin pregnancies, from a population of women who are predominantly at a higher risk of having a fetus with Down's syndrome (see prevalence for the population tested). Performances are dependant of laboratories fully reporting discordant results to Yourgene Health as they occur. Please note that the IONA® Nx NIPT Workflow is a Non-Invasive Prenatal Test (NIPT) based on analysis of cell-free fetal DNA (cffDNA) in maternal blood. From data held on file by Yourgene Health. Correct as of 31st Oct 2023.

SENSITIVITY AND SPECIFICITY

>99.9%

FAILURE RATE

ONLY 0.2%

Scan the QR code to access the latest performance data for the IONA® Nx NIPT Workflow following post-market surveillance

Technical Services

- Single point of call for installation, validation, training and ongoing technical services
- Installation Qualification (IQ) and Operational Qualification (OQ) coordination
- Bespoke workflow training
- Performance Qualification (PQ)

Ordering Information

Hardware and Software

Product Name	Part Code	Description
Yourgene® QS250 Instrument and Starter Kit	40110010	Yourgene® QS250 Instrument and starter kit. Includes USB cables and power supply. Starter kit Includes: One (1) Plate-based quantitation kit with sufficient labware, standards and fluorophores for the quantitation of up to 96 samples. PQ reagents: - Two (2) plates of DDLB with DNA ladder sample - Two (2) 2.0% size selection cassettes Camera calibration block
Yourgene®SP150 Liquid-handler Instrument	40110020	Includes the workstation and monitor. Please note that disposable tips must be purchased separately
NIPT Analysis Workstation	40140010	NIPT Analysis workstation pre-installed with NIPT Analysis Software
Yourgene Network-attached storage	40151000	External hardware to store data from the sequencer before being processed by analysis software (recommended)
Workflow Manager Workstation	40141000	Workstation to host the Yourgene Data Management System (DMS). Includes: Atlas Workflow Manager software, monitor, keyboard, mouse & mat, thermal label printer GK420t, high-performance 5095 resin thermal transfer ribbon, white polyester cryo labels and CR1400 code reader 1400

Reagents and Consumable Bundles

Product Name	Part Code	Description
IONA® Nx Manual Kit Bundle (exc ILMN seq kit)	NX-00-M01-Q01	Contains Yourgene cfDNA Extraction Kit, IONA® Nx cfDNA Library Prep Dx Kit and Ranger® Quantiselect Kit. Enough for 96 tests. Includes ILMN IP fee. Excludes ILMN Sequencing Kit
IONA® Nx Automated Kit Bundle (inc ILMN seq kit)	NX-01-S01-Q01	Contains Yourgene cfDNA Extraction Kit, IONA® Nx cfDNA Library Prep Dx Kit, Ranger® Quantiselect Kit, Yourgene® SP150 cfDNA Extraction Consumables and Yourgene® SP150 cfDNA Library Prep Consumables. Enough for 96 tests. Includes ILMN IP fee. Includes ILMN Mid-Output Sequencing Kits (x 2)
IONA® Nx Automated Kit Bundle (exc ILMN seq kit)	NX-00-S01-Q01	Contains Yourgene cfDNA Extraction Kit, IONA® Nx cfDNA Library Prep Dx Kit, Ranger® Quantiselect Kit, Yourgene® SP150 cfDNA Extraction Consumables and Yourgene® SP150 cfDNA Library Prep Consumables. Enough for 96 tests. Includes ILMN IP fee. Excludes ILMN Sequencing Kits
IONA® Nx Automated Kit Bundle for High-Throughput users (inc ILMN seq kit)	NX-01-S01-Q02	Contains Yourgene cfDNA Extraction Kit, IONA® Nx cfDNA Library Prep Dx Kit, Ranger® Quantiselect Kit, Yourgene SP150 cfDNA Extraction Consumables and Yourgene SP150 cfDNA Library Prep Consumables. Enough for 96 tests. Includes ILMN IP fee. Includes ILMN High-Output Sequencing Kit (x 1)

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