sagentia innovation

IONA® Nx

Yourgene Health

Bringing a non-invasive prenatal testing concept to life while satisfying Class III medical device requirements

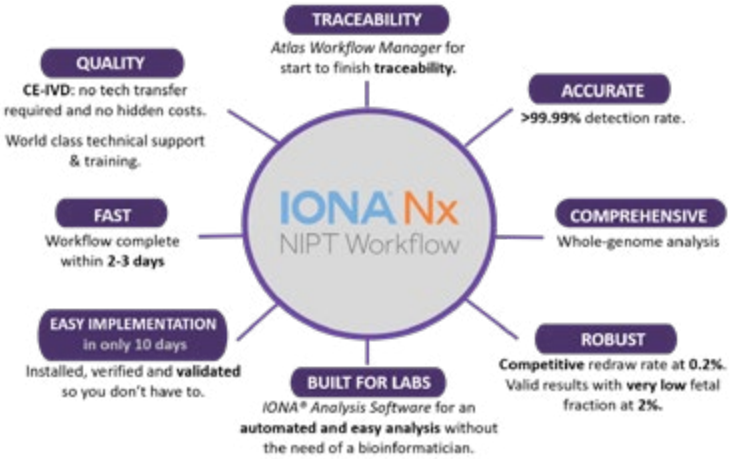
Yourgene Health had developed a bioinformatics method to diagnose three forms of foetal chromosomal trisomy: Down Syndrome, Edward Syndrome and Patau Syndrome. Using a maternal blood plasma sample rather than amniotic fluid, it eliminates the need for the invasive amniocentesis procedure traditionally associated with prenatal testing for genetic conditions. Molecular diagnostics specialist Yourgene Health sought technical support from Sagentia Innovation in the development of its IONA® Nx product.

Expertise

- C++ programming
- Data management
- Mathematical algorithms
- Software design

Domain knowledge

- Laboratory environment
- Test Operator needs
- Automated workflows
- CIII standard for high risk medical devices
- Medical grade software development
- Commercial insight and understanding



Yourgene Health

Our client asked:

The brief was to translate the mathematical algorithm at the heart of this non-invasive prenatal testing (NIPT) unit into a working diagnostic solution. The task was to build a functional software package to receive and analyse the DNA sequencer input, then detect any abnormalities related to chromosomes 13, 18 or 21. This had to be achieved in conformance with the CE medical device Class III (highest risk category) standard. The end goal was to ensure the software could integrate with various set-up configurations and third-party systems to provide fast and seamless foetal DNA analysis within an automated workflow.

The project story:

Our software engineers set about converting Yourgene Health's mathematical algorithm into a highly usable software application. It needed to receive the DNA sequence information and, through complex optimisation of the software, perform analysis within an acceptable timeframe. We selected, then adapted, two open-source algorithms to perform the heavy lifting at the core of the application: The Burrows-Wheeler Transform (BWT) data transformation algorithm is used to restructure the input data to make it more compressible. The dynamic Smith-Waterman Algorithm (SWA) is used to search the DNA sequences. Both algorithms were converted into the C++ programming language and optimised for use within the high performance, parallelised IONA® Nx workstation. The application also allows input of patient information, some of which relates to characteristics which are considered risk factors for chromosomal trisomy. This data serves as input to the algorithm, both manually and through an electronic HL7 interface from a lab information system, ensuring a high standard of workflow efficiency. We also configured the application ecosystem to enable seamless deployment of plugins and licences to Yourgene Health's customers.

Contact us

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Results: deliverables and outcomes

The IONA® Nx received CE marking with Class III medical device safety classification in June 2020. It launched in September 2020, positioned as 'the first CE marked IVD for prenatal screening enabling clinical laboratories around the world to establish their own quality-assured NIPT screening service'.

Furthermore, our efforts to streamline and automate the DNA analysis resulted in the time per test cycle being reduced from 8-12 hours to 2 hours, improving the efficiency and cost-effectiveness of the process.

These achievements mark an important milestone in Yourgene Health and Sagentia Innovation's ongoing and successful partnership. We are now working as a development partner on Yourgene Health's wider product ecosystem.

www.yourgene-health.com

> "Sagentia Innovation enabled us to accelerate the product development process thanks to a combination of technical and medical sector knowledge. Achieving the CE mark at our first attempt underlines the calibre of the team's expertise."