Contents

1. Introduction

This SOP describes the procedures to use the NIPT viewer to analyse samples and to manage the web application software.

1.1. Background

Illumina's NIPT VeriSeq assay produces two text files as output. The first file ("nipt_report") contains the final result with one row per sample. The second file ("supplementary_report") contains all measurements for each sample which has not failed quality control. Links to additional documentation on data format is provided in Section 11.

1.2. Purpose of procedure

This procedure will allow users to access and use the NIPT viewer and should be carried out when analysing all samples which have been processed through the VeriSeq assay. This procedure also includes admin instructions for maintaining the software which should be followed when interacting with the backend.

1.3. Scope

The first part of the procedure for using the app is intended to be followed by clinical scientists. The second part of the procedure for admin instructions is intended to be followed by members of the bioinformatics team.

1.4. Overview/summary of procedure

The app user instructions cover:

- Logging in to the app, registering an account and managing the user profile
- Analysing a worksheet
- Viewing file download history
- Getting help, and reporting an error or change request

The admin instructions include:

- Mapping required network drives
- Django database schema
- Deployment setup
- App admin page

2. Definitions

Term	Description
NIPT	Non-invasive prenatal testing
LLR	Log-likelihood ratio (probability of aneuploidy given fetal fraction estimate over the probability of no aneuploidy)
VM	Virtual machine

3. Responsibilities

Members of the bioinformatics team are responsible for system testing, data security, recording and reporting issues (e.g. errors, bug, system faults). Clinical scientists are responsible for using the web app only as described in this document and notifying the bioinformatics team of any issues that arise while following these steps.

4. External quality

Not applicable.

5. Version history

The code for the web application is stored in GitHub and any changes should be made in line with the WMRGL GitHub Code Review procedure and recorded as a new release, using semantic versioning.

Version	Date	Changes
v1.0.0-alpha	2020-04-22	Initial version ready for user testing

6. Software dependencies

Software	Version	Justification
Django	2.2.11	Web app framework (most recent long term supported version)
Pandas	1.0.1	Dataframe manipulation when parsing files
Plotly	4.5.4	Creating violin plots
Psycopg2	2.8.4	Connecting to PostgreSQL database

7. Reference materials

The application has been developed using test files and data format files supplied by Illumina on 26th March 2020 (Section 11).

8. Procedure

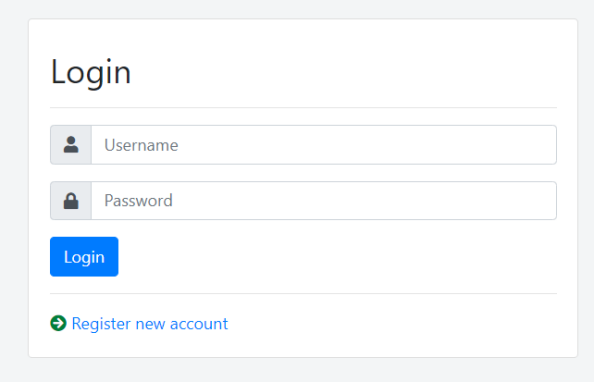
8.1. Web app user instructions

8.1.1. Accessing the NIPT viewer web app

The alpha version of the web app should only be opened with **Google Chrome** and can be accessed at the following address: http://10.147.33.228:8003/nipt.

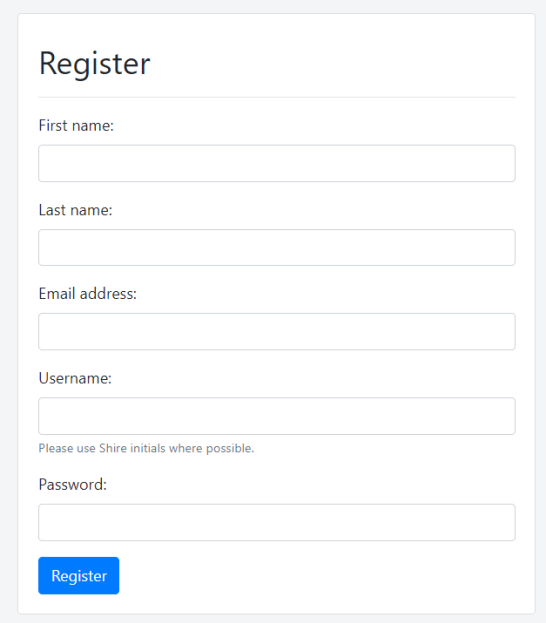
8.1.2. Logging in to the web app

If you have already registered, enter your username and password for this app and click “Login”. Otherwise, click on “Register new account” and move to next step.



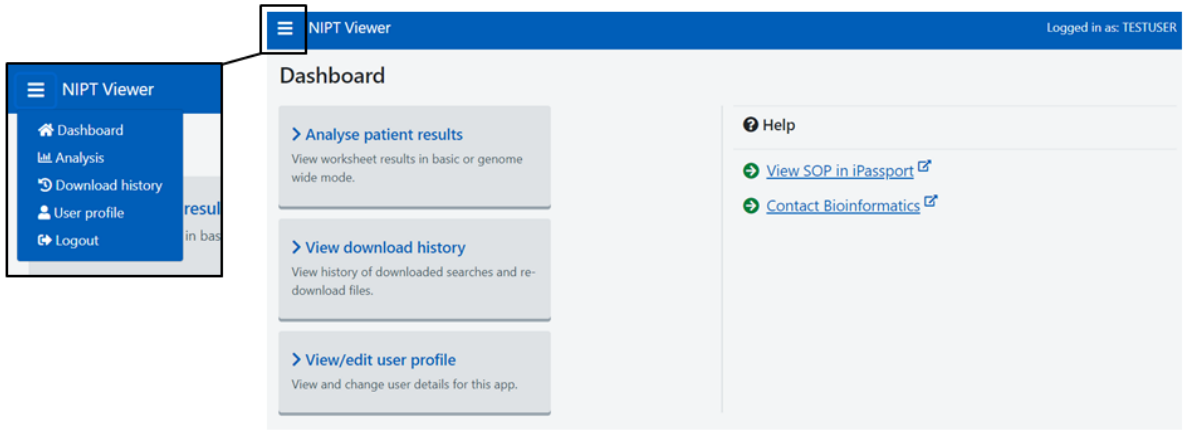
8.1.3. Registering a new account

Enter your details, using your trust email address and Shire initials as username if possible. Click “Register”. If successful, you will be logged in and redirected to the app home page.



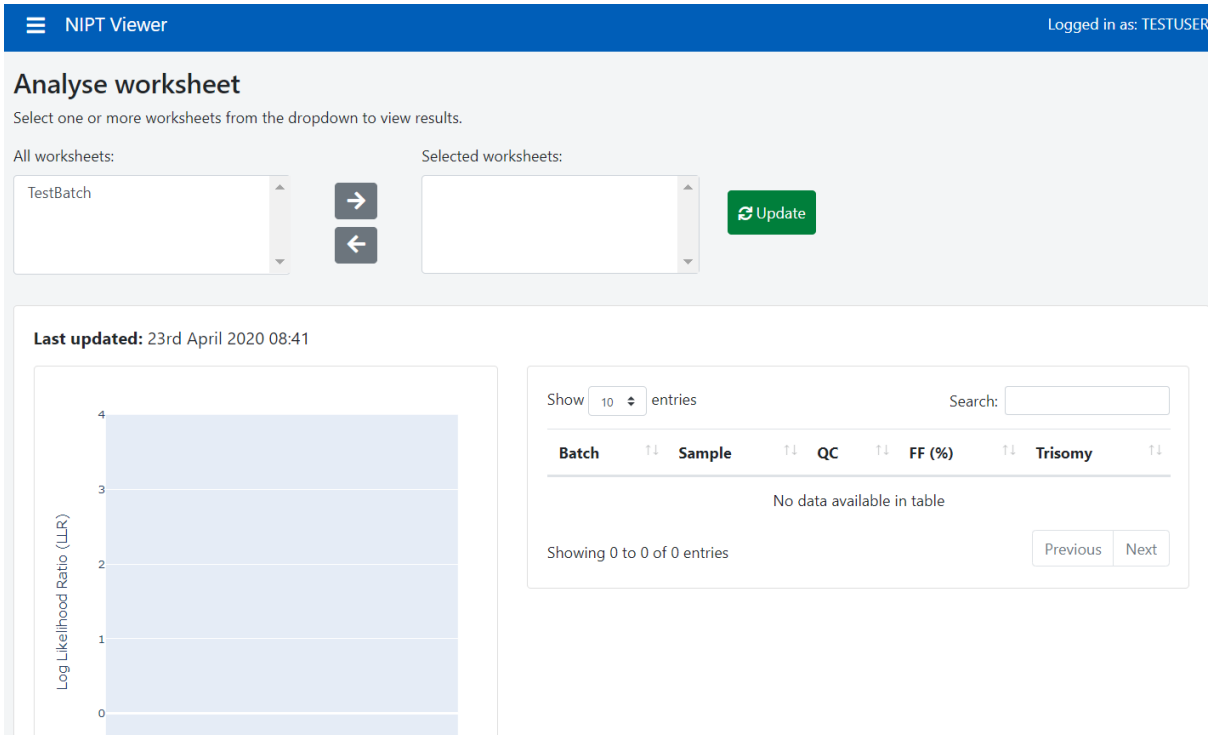
8.1.4. Home page

The home page dashboard provides links to the three main sections of the web app and help links to this SOP and to contact bioinformatics through email. The collapsed menu icon at the top left of the page can be used to navigate to the different pages.

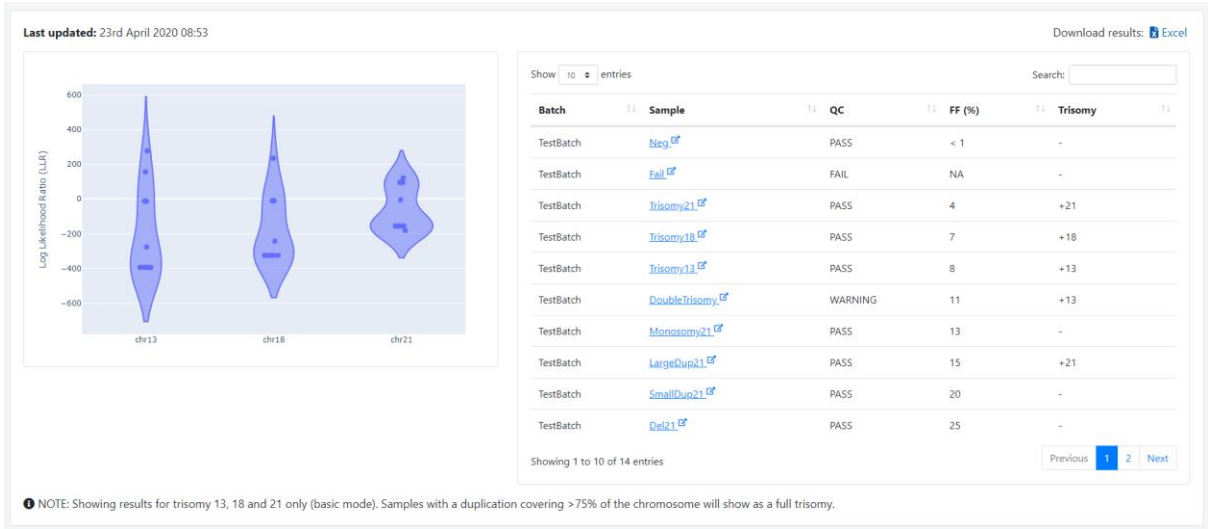


8.1.5. Analysing patient results

Select worksheets to view from the left box (multiple can be selected at once) and then click the right arrow. They will appear in the right box under “Selected worksheets”. Worksheets can be removed from being selected using the left arrow. When the worksheets have been selected, click “Update”. The results for all samples in the selected worksheet(s) will populate the table and plot.

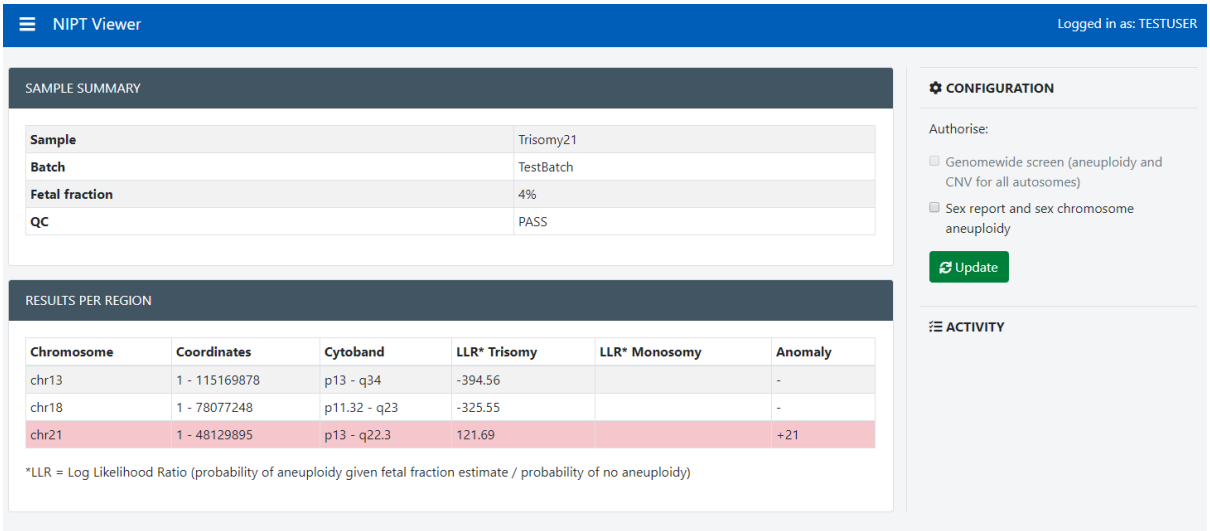


This page will only show results in **basic** mode, such that only T13, T18 and T21 will be shown and all other anomalies will initially be hidden in the app. In addition, samples run in **genomewide** mode with large duplications spanning >75% of chr13, chr18 or chr21 will be shown as a trisomy in this chromosome (this is the logic used by Illumina when running the assay in **basic** mode).



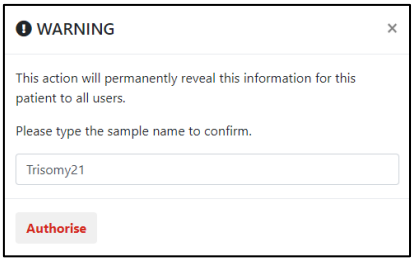
The table of results can be filtered using the “Search” box at the top right and ordered by clicking on the column headings. To download the current results, click on the Excel icon. This file will be saved

and the download recorded, viewable within the Download History page. The plot can also be manipulated using the cursor and icons at the top of the plot to aid analysis. To view a sample in more detail, click on the sample name in the table and this will open a new tab.

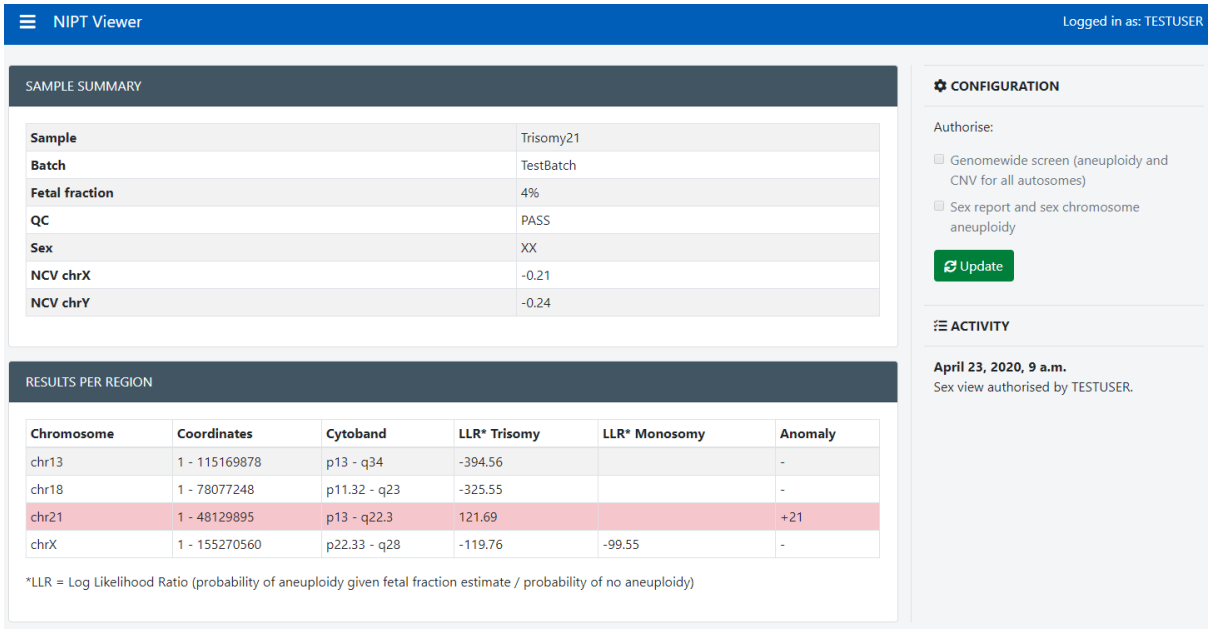


If a sample has not passed QC, there will be a message providing the reason for this. The results per region provide the LLR scores for trisomy and monosomy (only trisomy scores are shown while in basic mode). Results with anomalies will be highlighted in red.

This page can be configured to show the results in **genomewide** mode (all anomalies including trisomy, monosomy, duplications and deletions across all autosomes) and to show the sex results. The configurations available will depend on the original assay selection (for instance samples originally run in **basic** mode cannot be configured to show **genomewide** results). Select the desired checkboxes and click “Update”.

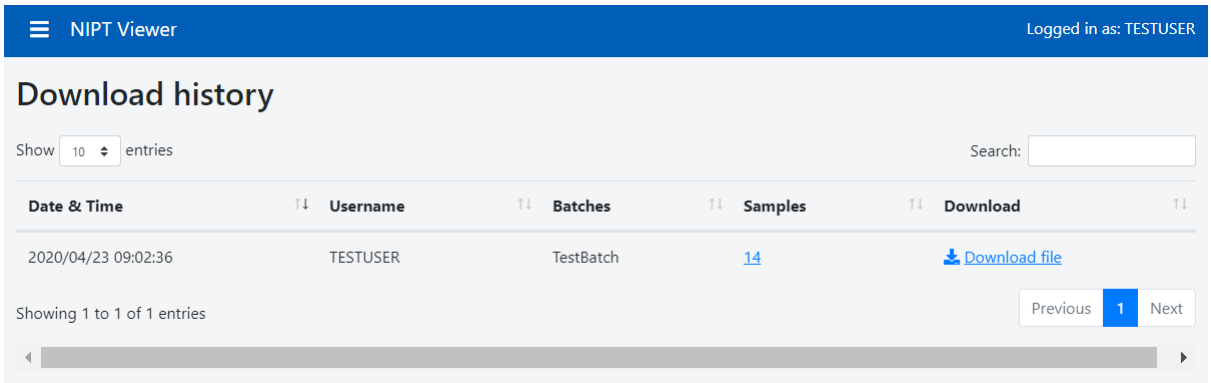


This warning message will then appear. To continue type in the sample name and click “Authorise”, otherwise close the message. Following authorisation, the results will be updated and a note of this action will be recorded under “Activity”.



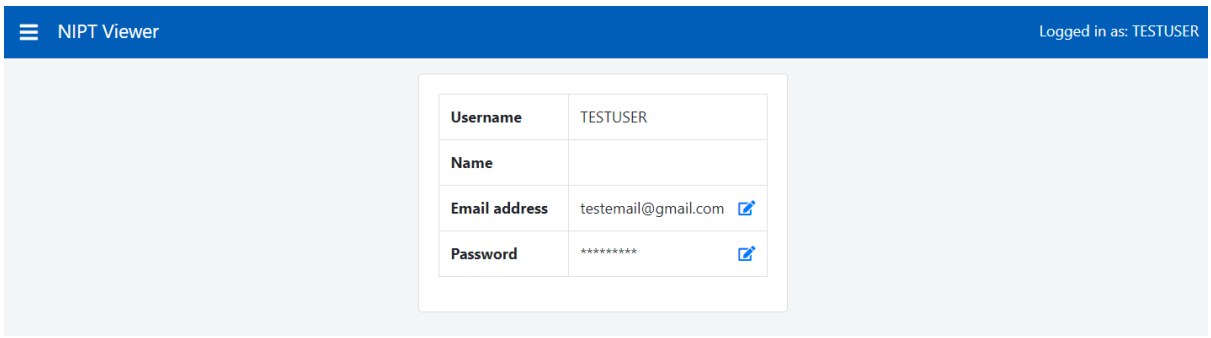
8.1.6 View download history

This page allows the user to view which searches have been downloaded or to retrieve a file previously created.



8.1.7. Manage user profile

Users can update their email or password using the user profile page. Click on the edit icon next to each row to update these.



8.2. Admin instructions for maintaining web app

8.2.1. Mapped network drives

The NIPT assay files are stored on the following drives:

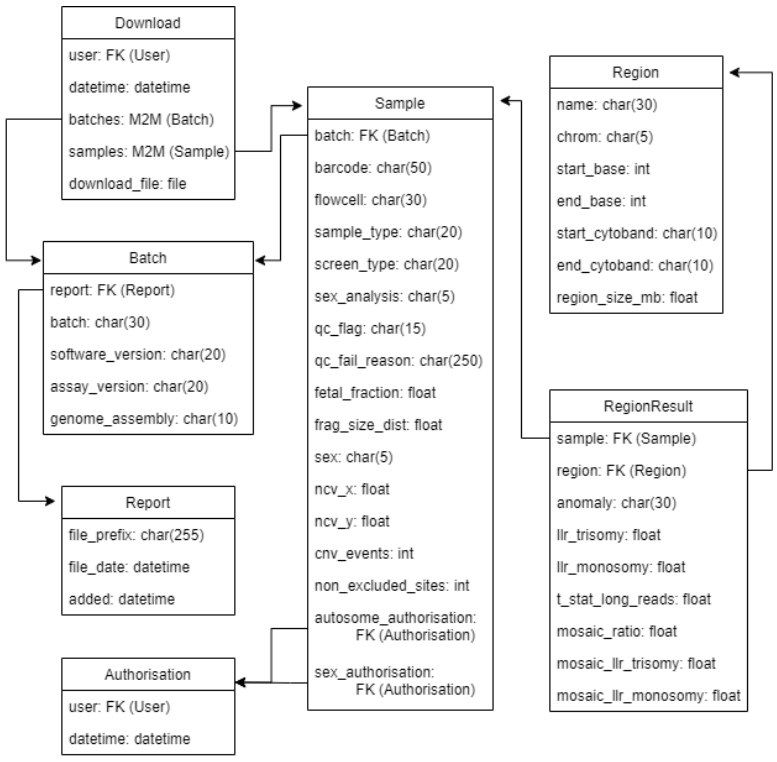
- 192.168.144.241\input (L:)
- 192.168.144.241\output (K:)

Username: sbsuser

Password: sbs123

8.2.2. Database schema

The database should be backed up automatically according to the Deployment of Django Web Applications to Bioinformatics Webserver procedure.



8.2.3. Deployment

The app is currently deployed on VM00 on port 8003. The app name is **nipt_viewer** with username **nipt_user**.

8.2.4. App admin page

The web app admin page is available at the following address: http://10.147.33.228:8003/admin. You must be a superuser to access this page.

9. Troubleshooting

Look at the Nginx and Gunicorn log files on the VM to determine the error.

10. Logging issues

Issues or change requests can be requested by emailing the Bioinformatics team or through GitHub repository issues page.

11. Links

Description	Link
Controlled documents	
VeriSeq laboratory procedure	?
Deployment of Django Web Applications to Bioinformatics Webserver	BI 01.01.22
WMRGL Github Code Review Procedure	BI 01.01.09
Code files	
GitHub repository	https://github.com/WMRGL/NIPT-Viewer
Change control	
Change request	CE 01.05.75
Useful references	
NIPT VeriSeq assay reference and test files	G:\Bioinformatics\NIPT VeriSeq documentation