

DEVELOPMENT OF INTEGRATED INFORMATION TECHNOLOGY (IT) SYSTEM FOR FAMILIAL HYPERCHOLESTEROLAEMIA SERVICES IN ENGLAND

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Introduction

- FH is an autosomal co-dominant condition that leads to high blood cholesterol from birth.
- PASS software developed for NHS Wales and adapted for NHS England.
- Purpose is to register FH patients and families and manage FH services, particularly family cascade testing.

NICE Quality Standard #5:

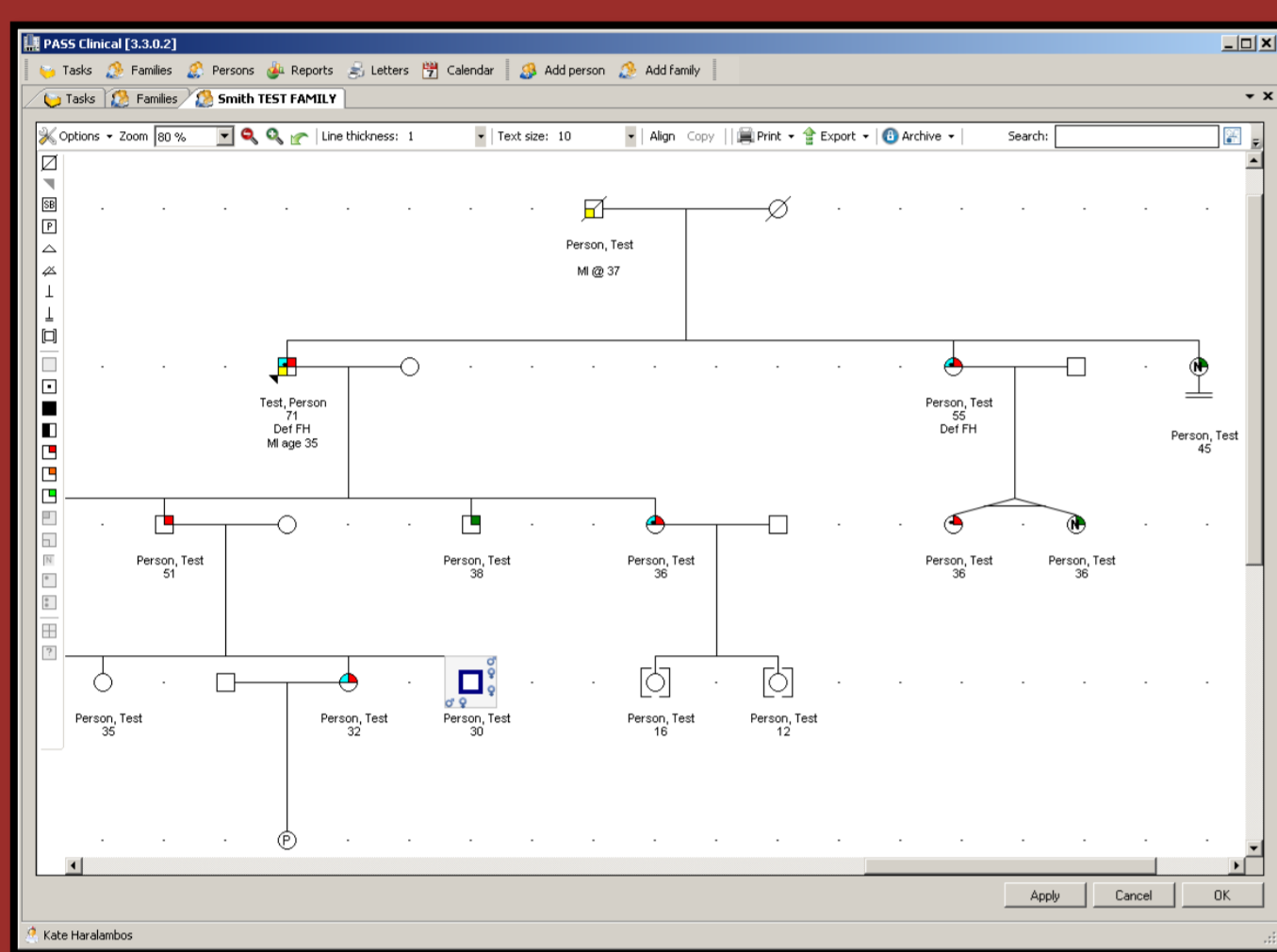
Relatives of people with a confirmed diagnosis of monogenic FH are offered DNA testing through a nationwide, systematic cascade process.

Key Features

- Pedigree drawing function
- Workflow management
- Templates and archiving
- Multisite working
- Audit, research & reporting

Local FH Service

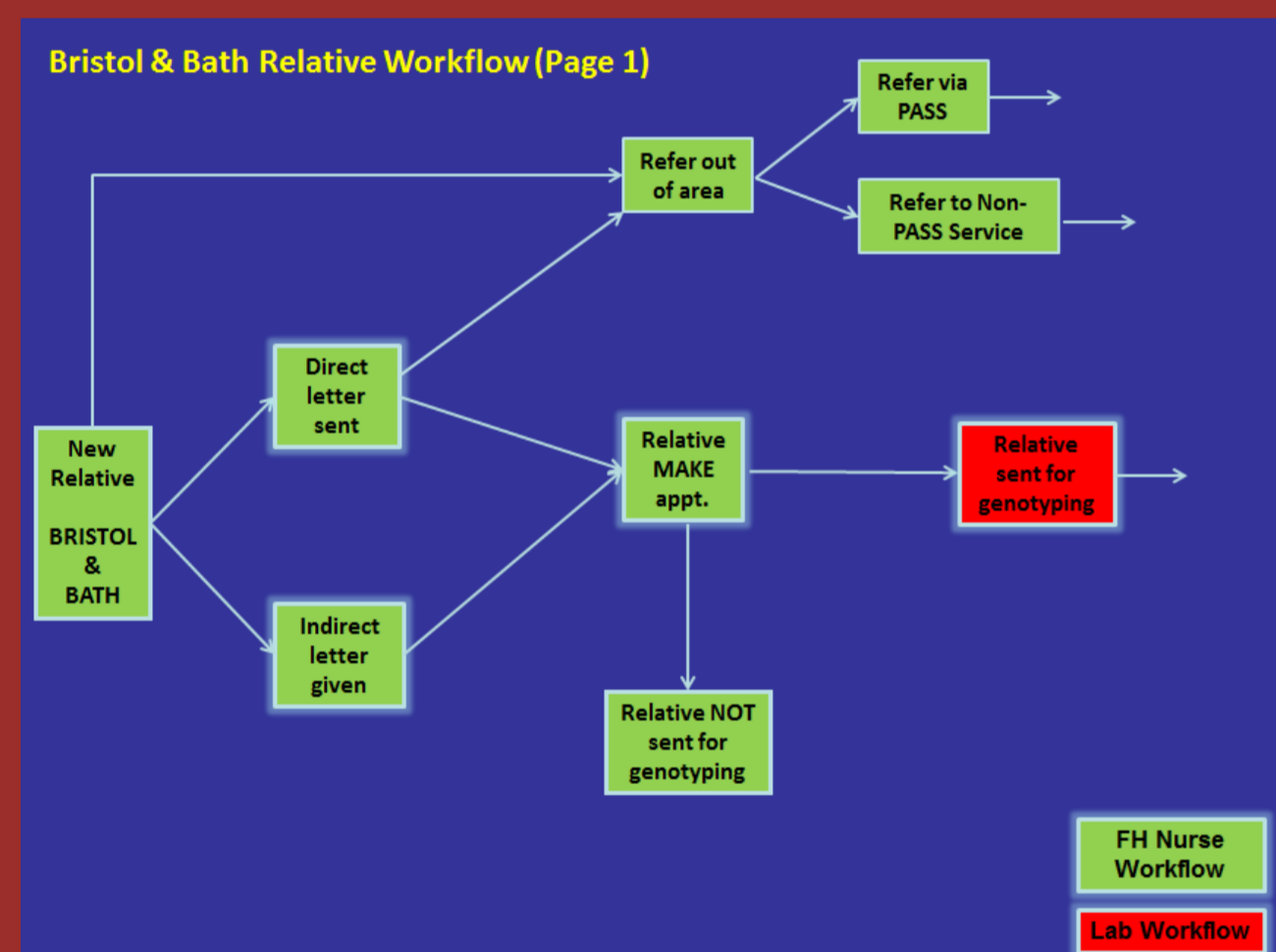
- FH patients and pedigrees registered locally.



Pedigrees can be created quickly and with ease, using a built in algorithm which automates drawing.

Visual markers and colour coding are used to show genetic status and phenotypical expression.

- Multidisciplinary workflow management

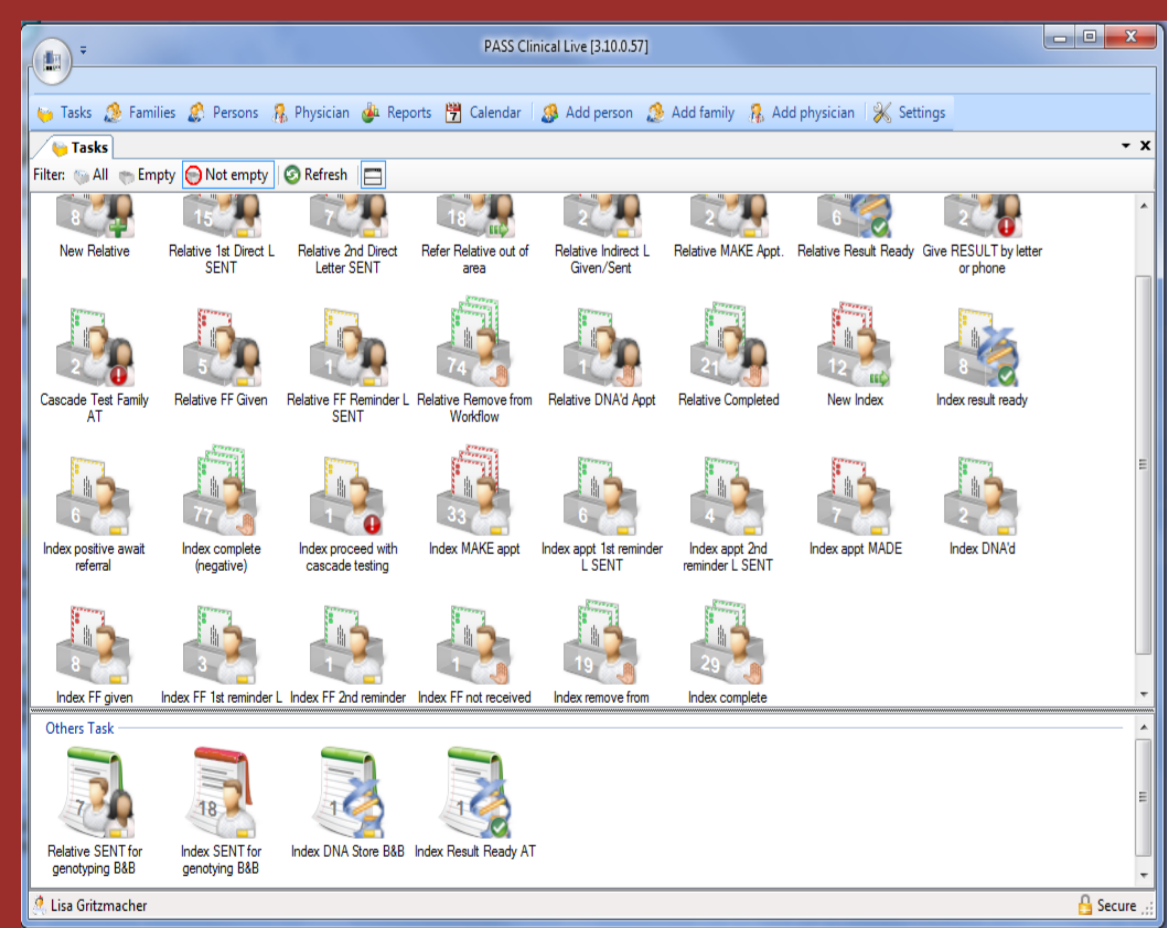


Allows systematic and multidisciplinary working via the workflows by integrating the workflow steps of each professional into one complete cascade testing workflow.

These can be flexible depending upon service design and professionals involved such as FH Nurses, Lipid consultants, administration and laboratory staff.

- The workflow visualised by a series of sequential boxes (user steps) and lists (co-workers steps).

- Patients entered into the workflow are located in one of these boxes/lists.



Allows the nurse to manage tasks such as letter creation and to manage and track their patient's progress through their care pathways.

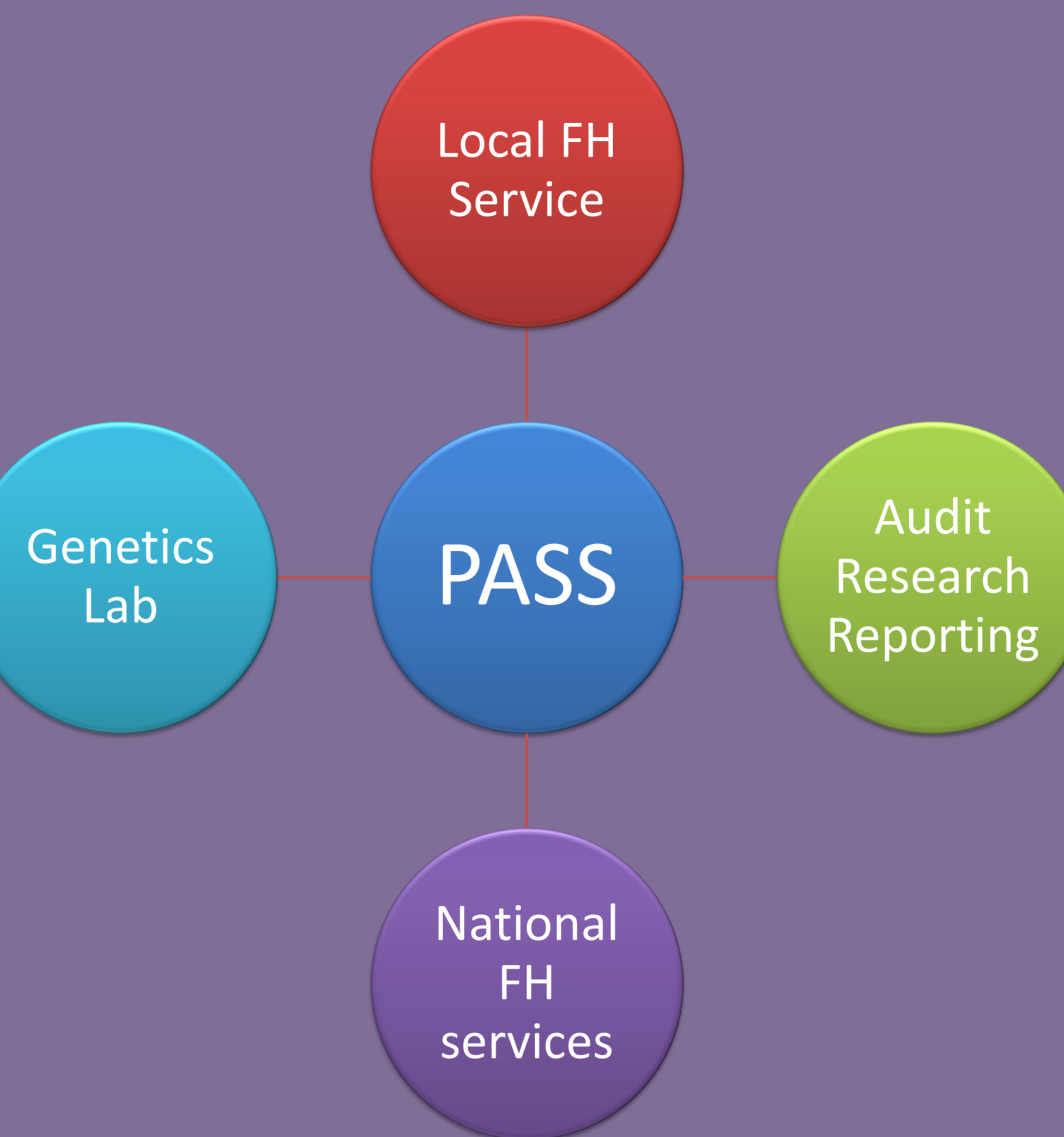
A "traffic light system" colour codes patients within workflow. If an individual has remained at a particular step over a pre-determined amount of time, they will be flagged amber or red, depending upon how overdue the particular task is.

- Workflows adaptable to different service designs.

- Allows dynamic development of workflows as a service develops.

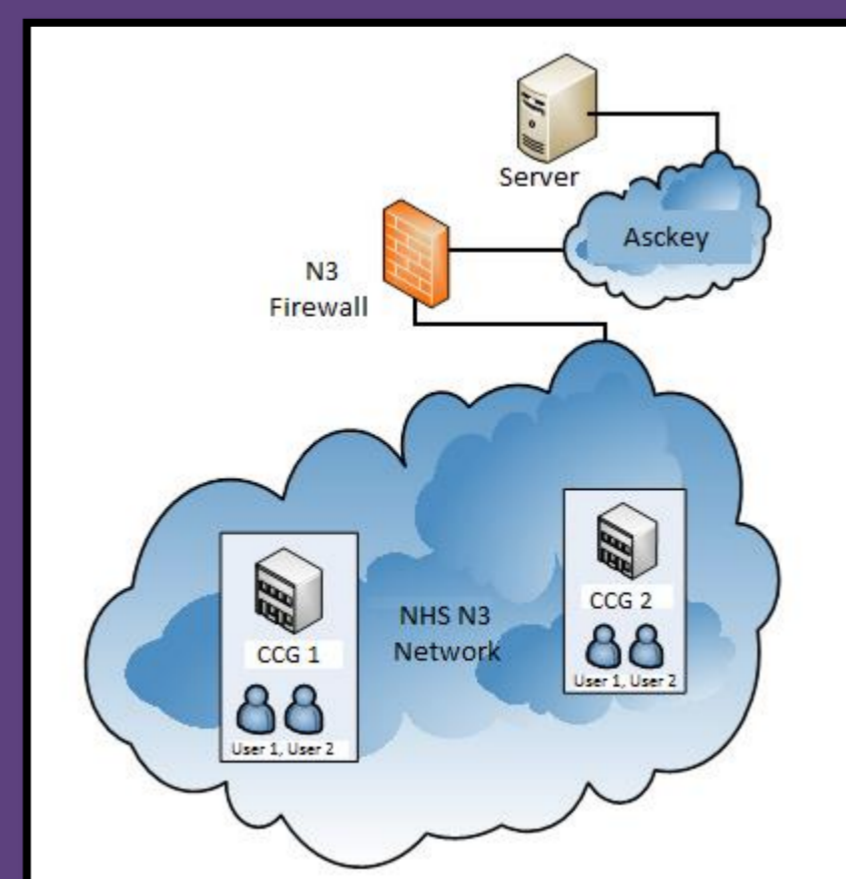
Genetic Laboratories

- Workflows can be integrated with genetic laboratories.
- DNA results/reports entered directly into PASS by Bristol genetics lab.
- Promotes good working relationships between lab and clinicians.



National FH Services

- Hosted on N3 network



- Coordinate testing at a National level

- PASS enables direct referral to other FH services across NHS England & allows coordination of cascade testing of a family by several different co-workers in different geographical locations.

- This facilitates a nationwide linked up service that aims for equity of diagnosis and treatment for all people with FH.

Audit, Research & Reporting

When used nationwide, the system standardises clinical and operational data collection between different centres and thus allows services to be evaluated locally and nationally.

Recruitment to research projects can be coordinated via the system, both by recording which studies patients are recruited to and creation of reports to aid recruitment, based on inclusion/exclusion criteria.

Patient Numbers

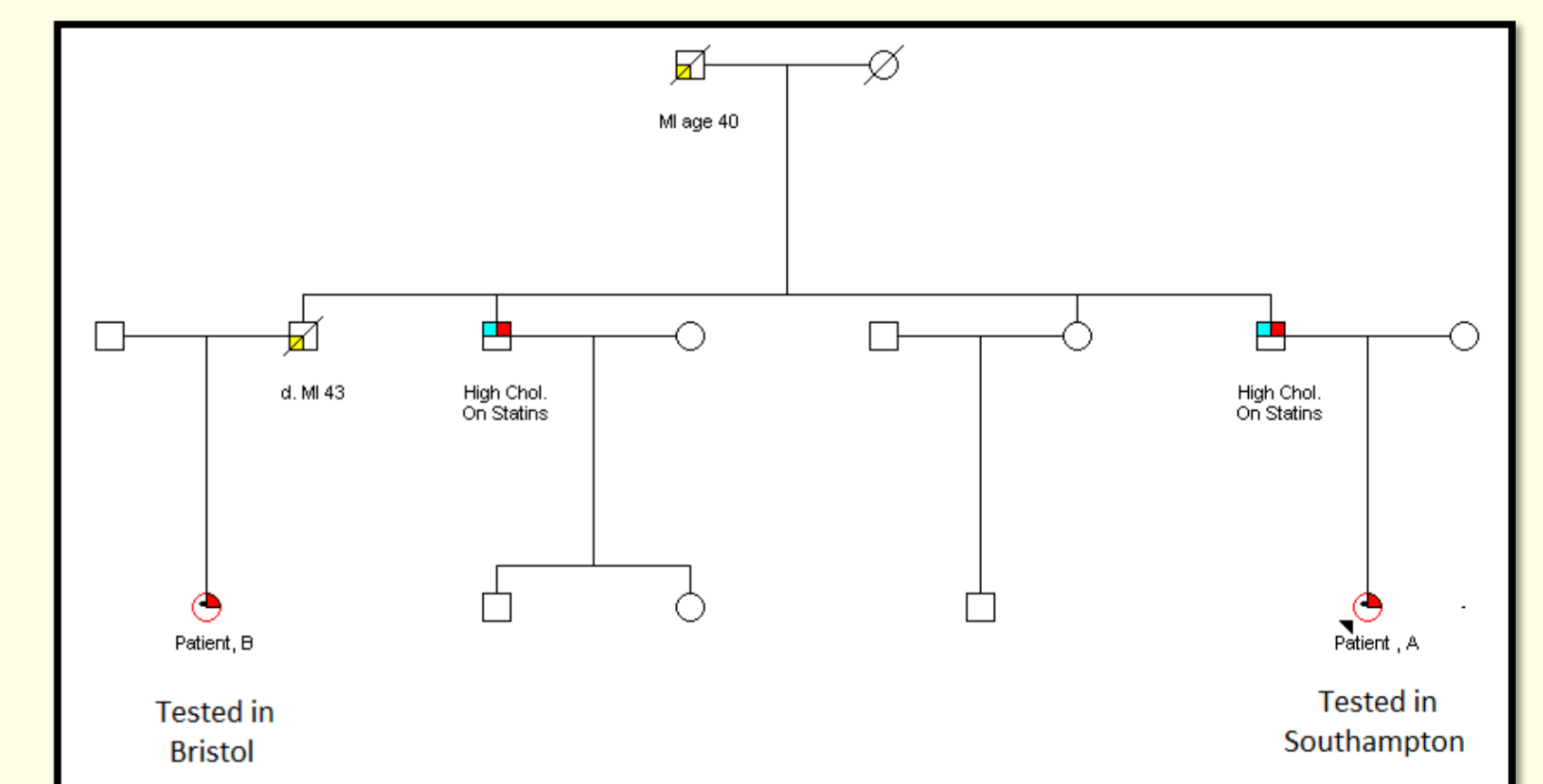
- 5455 in total (including potential at risk relatives)
- 1035 pedigrees
- 996 INDEXES with genetic results (587 positive & 409 negative)
- 444 RELATIVES with genetic result (281 positive & 163 negative)

Total number of patient in PASS with a genetic diagnosis: 868

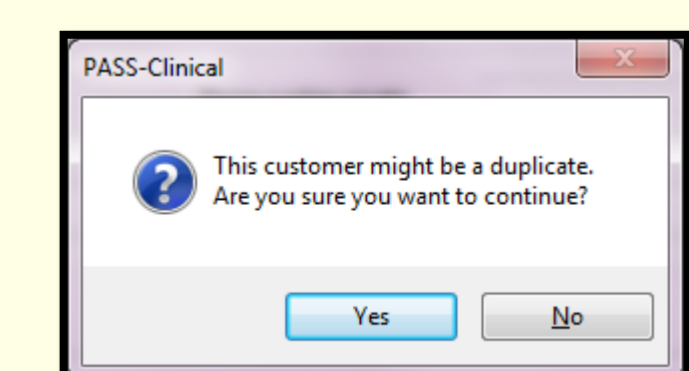
These figures are combined patient numbers from 7 different FH services in England.

Case Study – Linking Families

- Sample taken from index in Bristol (Patient B) & sent for genotyping.
- Pedigree drawn in PASS.



- PASS identified that Patient B was already registered in a family drawn by FH nurse in Southampton.



- Patient A (index in Southampton) was the cousin of Patient B and had already been sent for genotyping a month previously.
- Enabled linking of pedigrees & coordination between two FH services and genetics lab.
- Decision made to await results of Patient A, then if mutation identified, test Patient B as a cascade relative.
- Mutation identified in Patient A and also found to be inherited by Patient B.
- Prevented unnecessary and costly index genotyping, saving NHS resources by preventing duplication of testing.