

# **Technical Specification: Genetic Testing Data Submission**

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**Technical Specification for submission of the Genetic Testing data collection to the HSCIC**

**Clinical Indicators**

**Version 1.3**

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## Revision history

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<b>Version</b>	<b>Date</b>	<b>Summary of Changes</b>
1.0	August 2015	Published alongside Information Standards Notice
1.1	April 2016	Updates to information on Excel submission templates
1.2	1 <sup>st</sup> June 2016	Clarification to records that need to be flagged for molecular, page 6 bullet point 5 Clarification to Information on total numbers for submission; molecular submissions, page 6 bullet point 1
1.3	25 <sup>th</sup> July 2016	Clarification to Context following feedback. Correction to information on total numbers for submission, page 6 and page 10

## Context

This document contains details of the information being collected by the HSCIC from Genetic Testing laboratories in the UK.

In England this is being carried out on behalf of NHS England, under the “The Health and Social Care Information Centre (Establishment of Information Systems for NHS Services: Genetic Testing Data Collection) Directions 2014”. The burden of the collection has been assessed via the Standardisation Committee for Care Information (SCCI) and is the subject of an Information Standards Notice, reference: SCCI 2035.

The data is being collected on behalf of the Devolved Authorities of the UK by specific request of the respective Governments, as enabled under the terms of the Directions.

## Submission templates

There are two submission templates in use, one for molecular test activity and the other for cytogenetic test activity. These templates have been agreed by the UKGTN Clinical and Scientific Advisory Group.

The Excel files should be saved as Excel version 2007 or later (.xlsx).

They will be submitted by uploading the files via the HSCIC Secure Electronic File Transfer (SEFT) facility. Access to SEFT is granted to a named individual agreed with each lab. The HSCIC SEFT team contact these named individuals directly to set up the user name and password to provide this access. The software required for the use of SEFT is Java v1.6.0 or later.

If you are unsure who your named contact is, or have any questions about using SEFT, please contact [genetic.testing@hscic.gov.uk](mailto:genetic.testing@hscic.gov.uk)

The name of the lab to be used in submissions is that agreed for reporting purposes by the UKGTN Laboratory Membership and Audit working group. They are listed in Appendix 1.

File naming convention – please name your files consistently using the convention below

- CYT or MOL to identify file type
  - 4-character lab name (see Appendix 1)
  - Optional local ID – not mandatory but will assist if issues arise.
  - with .xlsx file extension
- e.g. 1 - CYT\_ABER\_localidV01.xlsx  
e.g. 2 - MOL\_ABER\_localidV02.xlsx

## Submission guidance – Molecular

### Instructions for MOLECULAR LABORATORIES to submit data to HSCIC for the review of genetic testing activity

#### General

- **Resident postcodes are required.** It is essential that the complete postcode is reported for each record. If possible please input the postcode as data items eight characters in length i.e. for 6 digit postcodes there are two spaces between each part of the code and for 7 digit postcodes there is only one space between each part of the code.
- If full resident postcodes are not available then the NHS/CHI number can be submitted.
- **Please submit Date of Birth and Postcode for all records.** If Date of Birth or Postcode is not available please submit the NHS/CHI number.

#### Submission criteria

- Laboratories are requested to only submit data on:
  1. The genetic test requests that they have analysed and reported on either **for local referrals or for imports minus the exclusions listed under “Exclusions for molecular” on page 5.**
  2. **Tests that are sent to non-UKGTN laboratories (based in UK or abroad) for NHS patients – that meet criteria 1 above**

**Laboratories should NOT send records where analysis was carried out in another UKGTN laboratory.** This will avoid double counting of ‘send-aways’ where testing is sent to other UKGTN labs while still allowing for the capture of data on tests that are sent to non-UKGTN laboratories.
- Molecular and cytogenetic laboratories that are part of a Regional Genetics Centre need to **ensure that data from the molecular and cytogenetic laboratories are submitted separately i.e. one Excel file for molecular records and one Excel file for cytogenetic records.**

- There is one **Excel file attached for completion for molecular records**. The Excel file has six worksheets:
  1. provides instructions for completion and is labelled “Instructions”
  2. is an empty data sheet for insertion of postcode/NHS/CHI number/date of birth/ data for tests requested from clinical genetics and is labelled “Genetic”
  3. is an empty data sheet for insertion of postcode/NHS/CHI number/date of birth data for tests requested from other specialties, excluding clinical genetics, and is labelled “Non-genetic”
  4. is an empty sheet for insertion of total number of activity and is labelled “Totals”.
  5. is a validation sheet which summarises the number of records which contain values which do not meet the expected format, and is labelled “validation report”
  6. is a comments sheet where users are asked to record any comments about validation errors, or any other issue which affects the submitted data, and is labelled “Comments”.
- There are named **exclusions** as detailed below.
- **Please flag specific conditions/techniques** as detailed below. If you are unable to flag these conditions/techniques please still submit all the data and advise that you were unable to flag the conditions/techniques listed. If unable to flag the conditions/techniques, please inform us which, if any, of the flagged conditions/techniques are tested at your laboratory.
- Please direct any queries to [genetic.testing@hscic.gov.uk](mailto:genetic.testing@hscic.gov.uk)

**Information to be provided for each test record completed by the laboratory for the data collection period**

Information required:

- 1) Year
- 2) Laboratory (only list your laboratory name)
- 3) Resident Postcode
- 4) NHS/CHI number (if postcode not collected)
- 5) Date of Birth

**Exclusions for molecular:**

- 1) Banking reports / DNA storage
- 2) Tests as part of research
- 3) tests for non UK residents
- 4) private referrals
- 5) Somatic / acquired molecular oncology tests (JAK2, KRAS, EGFR, BRAF MPL, FLT3, NPM1, SML, ALL etc) including MSI studies for HNPCC.
- 6) Thrombophilia tests (Factor V Leiden, Prothrombin)

- 7) Haemochromatosis tests
- 8) Haemophilia and Haemoglobinopathy (Sickle Cell and Thalassaemia) tests
- 9) Testing as part of population screening programme (e.g CF Newborn screening)
- 10) Pre- natal testing

### Records that need to be flagged for molecular:

- 1) Breast cancer – flagged with a 1 in the relevant column on the spreadsheet
- 2) Huntington Disease – flagged with a 2 in the relevant column on the spreadsheet
- 3) Fragile X – flagged with a 3 in the relevant column on the spreadsheet
- 4) molecular rapid aneuploidy tests – flagged with a 4 in the relevant column on the spreadsheet
- 5) arrayCGH and MLPA tests for the clinical purpose of analysing genomic imbalances / microdeletions only e.g sub telomeric deletions. Do not use this flag for tests where MLPA is used for single gene disorders e.g. inclusion of MLPA together with NGS or Sanger sequencing for BRCA1/2, Lynch syndrome, Rett syndrome, HSP etc nor where MLPA is used for conditions such as DMD, SMA, CMT, PWS or BWS.

### Split of data by specialty:

- Please provide data split between requests from clinical genetics specialty and requests from all other specialties. Please record referrals from clinical genetics specialty on the 'Genetic' worksheet and set 'Genetic flag' = G; record referrals from other specialties on the 'Non-genetic' worksheet and set 'Genetic flag' = 'NG'.

## Information on total numbers for submissions

### Molecular submissions

Please provide the following total numbers in the worksheet named 'Totals':

1. total number of **reports the laboratory issued** for all molecular analysis (**do not include storage and extractions**) where the laboratory carried out the analysis
2. total number of **reports the laboratory issued that met the submission criteria** where the laboratory carried out the analysis (excluding send-aways to UKGTN and non-UKGTN laboratories)
3. total number of cases that met the submission criteria that were sent to non-UKGTN member laboratories for analysis.
4. total number of cases that met the submission criteria that were sent to other UKGTN laboratories for analysis (i.e. send-aways)

**The total numbers requested are split between requests from clinical genetics and requests from other specialties.** The Excel worksheet named 'Totals' is the data sheet for recording of these total numbers.

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Please provide any known reasons for changes in genetic test provision since 2011/12, for example, any changes in local clinical practice which has significantly reduced or increased any particular test.

## Submission guidance – Cytogenetic

### Instructions for CYTOGENETIC LABORATORIES to submit data to HSCIC for the review of genetic testing activity

#### General

- **Resident postcodes are required.** It is essential that the complete postcode is reported for each record. If possible please input the postcode as data items eight characters in length i.e. for 6 digit postcodes there are two spaces between each part of the code and for 7 digit postcodes there is only one space between each part of the code.
- If full resident postcodes are not available then the NHS/CHI number can be submitted.
- **Please submit Date of Birth and Postcode for all records.** If Date of Birth or Postcode is not available please submit the NHS/CHI number.

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#### Submission criteria

- Laboratories are requested to only submit data on:
  1. The genetic test requests that they have analysed and **reported on either for local referrals or for imports for constitutional cases only AND minus the exclusions listed under “Exclusions for cytogenetic” on page 9.**
  2. Tests that are sent to non-UKGTN laboratories (based in UK or abroad) for NHS patients that meet criteria 1 above

**Laboratories should NOT send records where analysis was carried out in another UKGTN laboratory.** This will avoid double counting of ‘send-aways’ where testing is sent to other UKGTN labs while still allowing for the capture of data on tests that are sent to non-UKGTN laboratories.

- Molecular and cytogenetic laboratories that are part of a Regional Genetics Centre need to **ensure that data from the molecular and cytogenetic laboratories are submitted separately i.e. one Excel file for molecular records and one Excel file for cytogenetic records.**
- **Cytogenetics** laboratories should only submit data related to testing for **constitutional cases only.**
- There is one **Excel file attached for completion for cytogenetic records.** The Excel file has six worksheets:
  1. provides instructions for completion and labelled “Instructions”
  2. is an empty data sheet for insertion of postcode/NHS/CHI number/date of birth for tests requested from clinical genetics and is labelled “Genetic”
  3. is an empty data sheet for insertion of postcode/NHS/CHI number/date of birth for tests requested from other specialties, excluding clinical genetics, and is labelled “Non-genetic”
  4. is an empty sheet for insertion of total number of activity and is labelled “Totals”.
  5. is a validation sheet which summarises the number of records which contain values which do not meet the expected format, and is labelled “Validation Report”
  6. is a comments sheet where users are asked to record any comments about validation errors, or any other issue which affects the submitted data, and is labelled “Comments”.
- There are named **exclusions** as detailed below.
- The **cytogenetic labs are asked to flag records** as detailed below. If you are unable to flag these records please still submit all the data and advise that you were unable to flag them.
- Please direct any queries to [genetic.testing@hscic.gov.uk](mailto:genetic.testing@hscic.gov.uk)

**Information to be provided for each test record completed by the laboratory for the data collection period**

Information required:

- 1) Year
- 2) Laboratory (only list your laboratory name)
- 3) Resident Postcode
- 4) NHS/CHI number (if postcode not collected)
- 5) Date of birth

### **Exclusion for cytogenetics:**

- 1) Banking reports / DNA storage / culture & send-away
- 2) Tests as part of research
- 3) tests for non UK residents
- 4) private referrals
- 5) tests for acquired conditions
- 6) Testing as part of population screening programme (e.g CF Newborn screening)

### **Records that need to be flagged for cytogenetic:**

- 1) arrayCGH and MLPA tests for genomic imbalance / microdeletions – flagged with a 1 in the relevant column on the spreadsheet
- 2) All aneuploidy using QF-PCR and FISH – flagged with a 2 in the relevant column on the spreadsheet
- 3) Pre natal records – flagged with a 3 in the relevant column on the spreadsheet
- 4) Post natal records – flagged with a 4 in the relevant column on the spreadsheet

### **Split of data by specialty:**

- Please provide data split between requests from clinical genetics specialty and requests from all other specialties. Please record referrals from clinical genetics specialty on the 'Genetic' worksheet and set 'Genetic flag' = G; record referrals from other specialties on the 'Non-genetic' worksheet and set 'Genetic flag' = 'NG'.

## **Information on total numbers for submissions**

### **Cytogenetic submissions**

Please provide the following total numbers in the worksheet named 'Totals':

1. total number of **reports that laboratory issued** for all cytogenetic analysis (**do not include storage and extractions**) where the laboratory carried out the analysis
2. total number of reports the laboratory issued that met the submission criteria where the laboratory carried out the analysis (excluding send-aways to UKGTN and non-UKGTN laboratories)
3. total number of cases that met the submission criteria that were sent to non-UKGTN member laboratories for analysis.
4. total number of cases that met the submission criteria that were sent to other UKGTN laboratories for analysis (i.e. send-aways)

**The total numbers requested are split between requests from clinical genetics and requests from other specialties.**

The Excel worksheet named 'Totals' is the data sheet for recording of these total numbers.

## Appendix 1 – Laboratory names

List of laboratory names as agreed by the UKGTN Laboratory Membership and Audit working group for reporting purposes. (Four character codes for use in file submission are shown in brackets)

Aberdeen RGC (ABER)  
Belfast RGC (BELF)  
Birmingham IMD (BRMI)  
Birmingham RGC (BRMR)  
Bristol RGC (BRIS)  
Cambridge RGC (CAMB)  
Cardiff RGC (CARR)  
Dundee RGC (DUND)  
Edinburgh RGC (EDIN)  
Exeter RGC (EXET)  
Glasgow RGC (GLAS)  
Leeds RGC (LEED)  
Leicester RGC (LEIC)  
Liverpool RGC (LIVE)  
London Institute of Cancer Research (LICR)  
London Institute of Neurology (LINE)  
London King's Haemoglobinopathy (LKHA)  
London North East RGC GOSH (GOSH)  
London North West RGC KGC (LNWR)  
London Retinoblastoma (LRET)  
London South East RGC GSTT (GSTT)  
London South West RGC St Georges (STGE)  
London UCLH Biochemistry (UCLB)  
London UCLH Haemoglobinopathy (UCLH)  
Manchester RGC (MANC)  
Newcastle Mitochondrial Laboratory (NEWM)  
Newcastle RGC (NEWR)  
Nottingham RGC (NOTT)  
Oxford Haemoglobinopathy (OXFH)  
Oxford RGC (OXFR)  
Salisbury RGC (SALI)  
Sheffield RGC (SHEF)