Measure Number	Theme	Measure	Description & Provenance	Name of KPI	Numerator	Denominator	Period Type	Frequency	Data Source Numerator	Data Source Denominator	Data presentation	Target	Interpretation Guidance	Construction	Data Quality Concerns	Error Measurement	Notes_1	Notes_2	Notes_3
GEN01	Domain 2: Enhancing quality of life for people with long term conditions		Pick up rate for genetic testing	Proportion of tests that return a positive result for affected patients who have the test to determine a diagnosis and are seen in clinical genetics.	Number of tests that return a positive result ordered for symptomatic patients who have the test to determine a diagnosis and are seen in clinical genetics (exclude Fragile X and array CGH).	Number of tests ordered for symptomatic patients who have the test to determine a diagnosis for patients seen in clinical genetics (exclude Fragile X and array CGH).	Quarterly	Quarterly	Hospital clinical records		Trends to be monitored		Neutral	Numerator / denominator x 100			We will need to define the exclusions with the professional bodies.	Initially only exclusion is Fragile X testing and aCGH	a panel test counts as one test
GEN02a	Domain 3: Helping people to recover from episodes of ill health or following injury		Multi Disciplinary Clinics (MDC) - with patients in attendance - clinical genetics clinics	Proportion of clinical genetic clinics that are part of a MDC (not MDT)	Number of clinical genetic clinics that are part of a MDC (not MDT)	Total number of genetic clinics	Quarterly	Quarterly	Hospital activity data				Neutral	Numerator / denominator x 100			It is good practice to have MDC for some conditions that affect multiple body systems where a patient otherwise would need to have appointments to see different specialist clinicians. Agree that we need to establish baselines to determine a measurement range that would indicate that services are performing well against this indicator.	Only include clinics led, organised and commissioned by the commissioners of clinical genetics (do not include clinics organised by other specialties but attended by staff from clinical genetics). This will include NSCAG clinics.	
GEN02b	Domain 3: Helping people to recover from episodes of ill health or following injury		Multi Disciplinary Clinics (MDC) - with patients in attendance - where clinic has been organised by other specialties but attended by staff from Clinical Genetics	Rate of attendances by clinical genetics to clinics organised by other specialties	Number of clinics organised by other specialties but attended by staff from clinical genetics	Number of WTE clinical geneticists and genetic counsellors (excluding trainees)	Quarterly	Quarterly	Hospital activity data				Neutral	Numerator / denominator x 100			include clinics organised by other specialties but attended by staff from clinical genetics.		
GEN03	Domain 3: Helping people to recover from episodes of ill health or following injury		Clinical audits	Proportion of clinical audits completed and action plans put in place (the number and type of audits need to be agreed)	Number of clinical audits completed and action plans put in place (from the agreed list of audits)	Number of clinical audits that the clinical genetics department was expected to participate in	Annual (FY)	Annual (FY)	Dependent on specific audits agreed				Higher is better	Numerator / Denominator x 100			The number and type of clinical audits that services are expected to participate in will need to be agreed with the CQG/Lead Clinicians on an annual basis to establish how many audits they are expected to participate in.	For 2013/14 - Beckwith Wiedemann syndrome audit	
GEN04ai	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of Urgent Prenatal Rapid aneuploidy QF/PCR/FISH tests, Urgent Postnatal Blood Rapid aneuploidy tests and Urgent Haematology/Leukaemia Rapid PCR/FISH tests completed within 3 working days	Number in denominator completed within 3 working days	Number of Urgent Prenatal Rapid aneuploidy QF/PCR/FISH tests, Urgent Postnatal Blood Rapid aneuploidy tests and Urgent Haematology/Leukaemia Rapid PCR/FISH tests	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			Include all tests in the lab, not just those which are requested by clinical genetics.		
GEN04aii	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of Urgent Postnatal Blood Karyotype result tests completed within 10 calendar days	Number in denominator completed within 10 calendar days	Number of Urgent Postnatal Blood Karyotype result tests	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			Include all tests in the lab, not just those which are requested by clinical genetics.		
GEN04aiii	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of Urgent Prenatal Karyotype result tests and Urgent Haematology/Leukaemia Karyotype result tests completed within 14 calendar days	Number in denominator completed within 14 calendar days	Number of Urgent Prenatal Karyotype result tests and Urgent Haematology/Leukaemia Karyotype result tests	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / 100			Include all tests in the lab, not just those which are requested by clinical genetics.		
GEN04aiv	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of Routine Haematology/Leukaemia tests completed within 21 calendar days	Number in denominator completed within 21 calendar days	Number of Routine Haematology/Leukaemia tests	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			Include all tests in the lab, not just those which are requested by clinical genetics.		
GEN04av	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of Routine Postnatal Tests and Routine Chromosomal microarray and/or PCR/FISH tests for proband or parental samples requested after detection of an imbalance in proband completed within 28 calendar days	Number in denominator completed within 28 calendar days	Number of Routine Postnatal Tests and Routine Chromosomal microarray and/or PCR/FISH tests for proband or parental samples requested after detection of an imbalance in proband	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			Include all tests in the lab, not just those which are requested by clinical genetics.		
GEN04avi	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of Routine Chromosomal microarray tests for proband and parents referred together completed within 56 calendar days	Number in denominator completed within 56 calendar days	Number of Routine Chromosomal microarray tests for proband and parents referred together	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			Include all tests in the lab, not just those which are requested by clinical genetics.		
GEN04bi	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of PCR-based tests where the result is needed urgently for prenatal diagnosis completed within 3 working days	Number in denominator completed within 3 working days	Number of PCR-based tests where the result is needed urgently for prenatal diagnosis	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			Activation date is the date on which the sample (or all the required samples) and all information pertaining to the request is received by the testing laboratory. Reporting date is the date on which the result is first issued; by telephone, fax, e-mail or written report. The reporting time is the difference between the two dates measured in working days.	Include all tests in the lab, not just those which are requested by clinical genetics.	
GEN04bii	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of Southern blot tests where the result is needed urgently for prenatal diagnosis, PCR-based tests where the familial mutation is known, specific mutation tests, or gene tracking by microsatellite analysis completed within 2 weeks	Number of tests in denominator completed within 2 weeks (10 working days)	Number of tests where the result is needed urgently for prenatal diagnosis, PCR-based tests where the familial mutation is known, specific mutation tests, or gene tracking by microsatellite analysis	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			The 3 day/2 week reporting times for urgent tests assumes a minimum period of notification (suggest 5 working days; this could be included in a service specification). In addition that the familial mutation is known and/or informativeness of markers is established.	Include all tests in the lab, not just those which are requested by clinical genetics.	
GEN04biii	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of Non-urgent PCR-based tests where the familial mutation is known, specific mutation tests, or gene tracking by microsatellite analysis completed within 4 weeks	Number of tests in denominator completed within 4 weeks (20 working days)	Number of Non-urgent PCR-based tests where the familial mutation is known, specific mutation tests, or gene tracking by microsatellite analysis	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			Multiplex PCR tests to detect known mutations (eg CFTR ARMS PCR or DLA) are included within "specific mutation tests". A separate PCR test (eg. CFTR zygosity test for a non-p.Phe508del mutation detected by ARMS PCR) would have an additional 2 week reporting time. If samples are pre-screened for p.Phe508del prior to a multiplex PCR test, a single 2 (or 4) week reporting time applies.	Include all tests in the lab, not just those which are requested by clinical genetics.	
GEN04biv	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of Mutation screening or tests that require Southern blot analysis completed within 8 weeks	Number of tests in denominator completed within 8 weeks (40 working days)	Number of Mutation screening or tests that require Southern blot analysis	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			A normal Fragile X result would be reported within 2 (or 4) weeks (specific mutation test), but if Southern blotting was required to confirm an expansion mutation or a homozygous normal result in a female then an 8 week reporting time would apply for the entire test. This also applies to myotonic dystrophy and Huntington disease.		
GEN04bv	Domain 3: Helping people to recover from episodes of ill health or following injury		Laboratory reporting times	Proportion of next generation sequencing panels of >10 genes completed within 16 weeks	Number of tests in denominator completed within 16 weeks (80 working days)	Number of next generation sequencing panels of >10 genes	Quarterly	Quarterly	Laboratory reports				Higher is better	Numerator / Denominator x 100			Temporary category to account for longer reporting times during implementation of new technology	Include all tests in the lab, not just those which are requested by clinical genetics.	
GEN05	Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm		Educational sessions provided by clinical genetics to other specialties to support genetics in mainstream medicine	Rate of educational sessions provided by clinical genetics to other specialties	Number of educational sessions provided by clinical genetics to other specialties	Number of WTE clinical geneticists and genetic counsellors (excluding trainees)	Quarterly	Quarterly	Hospital records				There is a need to assess average number of educational sessions in time period that would be expected as good practice in order to set a range for interpretation guidance.				An educational session is a formal post graduate education session lasting an an hour (excludes undergraduate teaching)		
GEN06	Domain 4: Ensuring that people have a positive experience of care		Poor patient experience	Rate of written complaints about the genetics department	Number of written complaints about the genetics department received during period	Total number of patient contacts (appointments including telephone contacts that replace a face-to-face appointment) in period	Quarterly	Quarterly	Hospital records				Lower is better	Numerator / Denominator x 100					
GEN07	Domain 4: Ensuring that people have a positive experience of care		Good patient experience	Rate of letters/emails from patients, carers or non-genetics consultants registering thanks to the genetics department	Number of letters/emails from patients, carers or non-genetics consultants registering thanks to the genetics department received during period	Total number of patient contacts (appointments including telephone contacts that replace a face-to-face appointment) in period	Quarterly	Quarterly	Hospital records				Higher is better	Numerator / Denominator x 100					
GEN08	Domain 4: Ensuring that people have a positive experience of care		Patients waiting excessively for pre-natal (PN) genetic test results where the patient is seen in the clinical genetics department	Proportion of patients receiving test result within 5 working days after the clinic recieves the laboratory report for PN genetic test results	Of all patients seen in clinical genetics who had prenatal diagnosis during the period, the number who received their prenatal genetic test result within 5 working days of the clinic receiving the laboratory report.	Number of patients seen in clinical genetics who had prenatal diagnosis during period	Quarterly	Quarterly	Hospital clinical records				Higher is better	Numerator / Denominator x 100			In clinical genetics (unless patient choice has opted to recieve results in the post) in most cases, the results should be communicated back to the family members face-to-face and more likely for PN test results this is done over the phone by a consultant geneticist or genetic counsellor. This is because unlike other biochemical and pathology blood tests, genetic test results have lots of implications either way for family members and this all needs to be explained. Good practice would advocate seeing or consultant calling patients to give them results. We suggest that this could be measured by reviewing patient appointments attended or phone calls made to explaine(receive) genetic test results. There would need to be in place the ability to code clinical records for appointments attended or phone calls made to recieve genetic test results.		
GEN09	Domain 4: Ensuring that people have a positive experience of care		Do Not Attends (DNA) as defined in the Data Dictionary (http://www.datadictionary.nhs.uk/data_dictionary/attributes/a/an/attended_or_did_not_attend_de.asp?shownav=1)	Proportion of appointments that are not attended	Number of DNAs within period	Total number of appointments booked during period within clinical genetics (311 treatment function). This can be taken as the total number recorded on PAS including DNAs	Quarterly	Quarterly	Hospital clinical records				Lower is better	Numerator / Denominator x 100					
GEN10	Domain 4: Ensuring that people have a positive experience of care		Patients counselled without a referral	Rate of patients counselled without a referral	Number of patients counselled without a referral during period	Total number of patients attending appointments during period	Quarterly	Quarterly	Hospital clinical records				Lower is better	Numerator / Denominator x 100			Trends need to be monitored		
GEN11a	Domain 4: Ensuring that people have a positive experience of care		Patients co-counselled by a genetic counsellor and doctor during period	Rate of patients co-counselled by a genetic counsellor and doctor during period	The number of clinic appointments where a genetic counsellor co-counsels with a doctor (excluding MDC co-counselling activity)	Total number of clinical genetics appointments of all types during period (appointments including telephone contacts that replace a face-to-face appointment)	Quarterly	Quarterly	Hospital clinical records				TBC		It is recognised that hospital IT systems may not record counsellor non-consultant appointments therefore this measure is intended to promote better recording of these appointments.		Ideally this would be an average of rates measured per patient but it's recognised that at present it is unlikely that trusts would be able to measure this.		
GEN11b	Domain 4: Ensuring that people have a positive experience of care		Patients counselled independently by a genetic counsellor	Rate of appointments counselled independently by a genetic counsellor during period	The number of independent genetic counsellor clinic appointments (excluding planned pre-clinic appointments preparing for a doctor's appointment)	Total number of clinical genetics appointments of all types during period (appointments including telephone contacts that replace a face-to-face appointment)	Quarterly	Quarterly	Hospital clinical records				TBC		It is recognised that hospital IT systems may not record counsellor non-consultant appointments therefore this measure is intended to promote better recording of these appointments.		Ideally this would be an average of rates measured per patient but it's recognised that at present it is unlikely that trusts would be able to measure this.		
GEN11c	Domain 4: Ensuring that people have a positive experience of care		Patient appointments held in preparation for a doctor's appointment	Rate of genetic counsellor clinic appointments held in preparation for a doctor's appointment	The number of genetic counsellor clinic appointments held in preparation for a subsequent doctor's appointment	Total number of clinical genetics appointments of all types during period (appointments including telephone contacts that replace a face-to-face appointment)	Quarterly	Quarterly	Hospital clinical records				TBC		It is recognised that hospital IT systems may not record counsellor non-consultant appointments therefore this measure is intended to promote better recording of these appointments.		Ideally this would be an average of rates measured per patient but it's recognised that at present it is unlikely that trusts would be able to measure this.		
GEN12a	Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm		Serious Untoward Incidents regarding patient care	Number of serious Untoward Incidents involving patient care	Number of serious Untoward Incidents involving patient care	N/A	Quarterly	Quarterly	SUI reporting system								Suggest we review against patient numbers seen / number of lab reports - depending on the incident.		
GEN12b	Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm		Serious untoward Incidents regarding lab tests	Rate of serious Untoward Incidents involving lab tests	Number of serious Untoward Incidents involving lab tests	Number of lab tests during period	Quarterly	Quarterly	SUI reporting system								Suggest we review against patient numbers seen / number of lab reports - depending on the incident.		
GEN13	Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm		External Quality Assurance	Episodes of poor performance in EQA schemes the laboratory participates in	Episodes of poor performance notified to the laboratory by UKNEQAS, EMQN or another EQA service provider	Number of EQA schemes participated in	Annual (FY)	Annual (FY)	EQA								It is mandatory that all labs participate in at least one EQA scheme. The EQA schemes are test specific by condition and therefore we would expect the specific schemes to vary by lab as different labs across the country provide different tests for different conditions.		
GEN14	Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm		Activity audits (laboratory)	Proportion of audits participated in by the genetics laboratory (the number and type of audits need to be agreed)	Number of audits (from the agreed list of audits) that genetics laboratory participates in	Number of audits that the genetics laboratories should be participating in	Annual (FY)	Annual (FY)	Dependent on specific audits agreed								As per Clinical Molecular Genetics Association for Clinical Cytogenetics and UKGTN audits requested.	to include workforce audit	
GEN15	Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm		Non-Adherence to UKGTN Testing Criteria (laboratory) as per UK GTN website.	Proportion of test requests from clinical genetics that did not comply to UKGTN Testing Criteria where TC apply.	Number of tests requested by clinical genetics from specified list that did not comply with UKGTN Testing Criteria	Number of tests requested from specified list	Quarterly	Quarterly	clinical records				Lower is better				All UKGTN Testing Criteria are available from the UKGTN website. The 3 tests for which adherence to UKGTN testing criteria are: 1) Silver Russell Syndrome, 2) Kabuki syndrome, 3) MEN2	Review after Q1 submission	