Measu	Theme	Measure Description & Provenance	Name of KPI	Numerator	Denominator	Period Type	Frequency	Data Source	Data Source	Data 1	Target 1	Interpretation Guidance	Construction	Data Quality Error	Notes_1	Notes_2	Notes_3
Numbe GEN01	Domain 2: Enhancing quality of life	Pick up rate for genetic testing	Proportion of tests that return a positive	Number of tests that return a positive result ordered for	Number of tests ordered for symptomatic patients who have the test to determine a diagnosis for patients seen in clinical genetics	Quarterly	Quarterly	Numerator Hospital clinical	Denominator Trends to be	presentation	N	Veutral	Numerator / denominator	Concerns Measurem	We will need to define the exclusions with the professional bodies.	Initially only exclusion is Fragile X testing and	a panel test counts as one
	for people with long term conditions		result for affected patients who have the test to determine a diagnosis and are seen in clinical genetics.	symptomatic patients who have the test to determine a diagnosis and are seen in clinical genetics (exclude Fragile X and array CGH).	test to determine a diagnosis for patients seen in clinical genetics (exclude Fragile X and array CGH).			records	monitored			:	x 100			aCGH	test
GEN02a	Domain 3: Helping people to recover from episodes of ill health	Multi Disciplinary Clinics (MDC) - with patients in attendance -		Number of clinical genetic clinics that are part of a MDC (not MDT)	Total number of genetic clinics	Quarterly	Quarterly	Hospital activity data			N	Veutral	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	Only include clinics led, organised and commissioned by the commissioners of clinical	
	or following injury	clinical genetics clinics		· · · ·											different specialist clinicians. Agree that we need to establish baselines to determine a measurement range that would indicate that services are performing	genetics (do not include clinics organised by other specialties but attended by staff from	
															well against this indicator.	clinical genetics). This will include NSCAG clinics.	
GEN02b	Domain 3: Helping people to recover from episodes of ill health	with patients in attendance -	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			N	Veutral :	Numerator / denominator x 100		include clinics organised by other specialties but attended by staff from clinical genetics.		
	or following injury	where clinic has been organised by other specialties but attended															
C (1903)	An and A total and A total	by staff from Clinical Genetics Clinical audits	Proportion of clinical audits completed and									and the first state of the stat					
GENUS	Domain 3: Helping people to recover from episodes of ill health or following injury	Clinical audits	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)	specific audits				ligher 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 CGS/Lead Clinicians on an annual basis to establis how many sudits they are expected to participate in.	For 2013/14 - Beckwith Wiedemann syndrome h audit	
GEN04ai	Domain 3: Helping people to	Laboratory reporting times	Proportion of Urgent Prenatal Rapid	Number in denominator completed within 3 working	Number of Urgent Prenatal Rapid aneuploidy QF/PCR/FISH tests,	Quartedy	Quarterly	Laboratory reports			н	ligher is better	Numerator / Denominator		Indude all tests in the lab, not just those which are requested by clinical genetics.		
	recover from episodes of ill health or following injury		aneuploidy QF/PCR/FISH tests, Urgent Postnatal Blood Rapid aneuploidy tests and	days	Urgent Postnatal Blood Rapid aneuploidy tests and Urgent Haematology/Leukaemia Rapid PCR/FISH tests		,						x 100		,, _,, _		
			Urgent Haematology/Leukaemia Rapid PCR/FISH tests completed within 3 working														
GEN04aii	Domain 3: Helping people to	Laboratory reporting times	days Proportion of Urgent Postnatal Blood	Number in denominator completed within 10 calendar	Number of Urgent Postnatal Blood Karyotype result tests	Quarterly	Quarterly	Laboratory reports	5		н	ligher is better	Numerator / Denominator		Include all tests in the lab, not just those which are requested by clinical genetics.		
	recover from episodes of ill health or following injury		Karyotype result tests completed within 10 calendar days Proportion of Urzent Prenatal Karyotype	days									x 100				
GEN04alli	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	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	5		H	ligher is better	Numerator / Denominator x 100		Include all tests in the lab, not just those which are requested by clinical genetics.		
	or ronowing injury		tests completed within 14 calendar days														
GEN04aiv	Domain 3: Helping people to recover from episodes of ill health	Laboratory reporting times	Proportion of Routine Haematology/Leukaemia tests completed	Number in denominator completed within 21 calendar days	Number of Routine Haematology/Leukaemia tests	Quarterly	Quarterly	Laboratory reports	5		н	figher is better	Numerator / Denominator x 100		Include all tests in the lab, not just those which are requested by clinical genetics.		
GEN04av	or following injury Domain 3: Helping people to	Laboratory reporting times	within 21 calendar days Proportion of Routine Postnatal Tests and	Number in denominator completed within 28 calendar		Quarterly	Quarterly	Laboratory reports	5		н	ligher is better	Numerator / Denominator		Include all tests in the lab, not just those which are requested by clinical genetics.		
	recover from episodes of ill health or following injury		Routine Chromosomal microarray and/or PCR/FISH tests for proband or parental	days	microarray and/or PCR/FISH tests for proband or parental samples requested after detection of an imbalance in proband							:	x 100				
			samples requested after detection of an imbalance in proband completed within 28 calendar days														
GEN04avi	Domain 3: Helping people to recover from episodes of ill health	Laboratory reporting times	Proportion of Routine Chromosomal microarray tests for proband and parents	Number in denominator completed within 56 calendar	Number of Routine Chromosomal microarray tests for proband and parents referred together	Quarterly	Quarterly	Laboratory reports	5		н	figher is better	Numerator / Denominator x 100		Include all tests in the lab, not just those which are requested by clinical genetics.		
	or following injury		referred together completed within 56 calendar days	P									~				
GEN04bi	Domain 3: Helping people to recover from episodes of ill health	Laboratory reporting times	Proportion of PCR-based tests where the result is needed urgently for prenatal	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	5		н	figher 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.	Include all tests in the lab, not just those which are requested by clinical genetics.	
	or following injury		diagnosis completed within 3 working days												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 date	5	
GEN04bii	Domain 3: Helping people to	Laboratory reporting times				Quarterly	Quarterly	Laboratory reports	s		н	figher is better	Numerator / Denominator		measured in working days. The 3 day/2 week reporting times for urgent tests assumes a minimum period of	Include all tests in the lab, not just those which are	
	recover from episodes of ill health or following injury		result is needed urgently for prenatal diagnosis, PCR-based tests where the familial	weeks (10 working days)	urgently for prenatal diagnosis, PCR-based tests where the familial mutation is known, specific mutation tests, or gene								x 100		notification (suggest 5 working days; this could be included in a service specification). In addition that the familial mutation is known and/or	requested by clinical genetics.	
			mutation is known, specific mutation tests, o gene tracking by microsatellite analysis	r	tracking by microsatellite analysis										informativeness of markers is established.		
	An and A total and A total	i de ser en este al ser el ser	completed within 2 weeks			0						and the first state of the stat					
GEN04bii	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, specifi mutation tests, or gene tracking by		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	1		H	ligher is better	Numerator / Denominator x 100		Multiplex PCR tests to detect known mutations (eg CFTR ARMS PCR or OLA) 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	Include all tests in the lab, not just those which are requested by clinical genetics.	
	or following injury		microsatellite analysis completed within 4 weeks		Inclosatence analysis										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.		
GEN04biv	Domain 3: Helping people to	Laboratory reporting times	Proportion of Mutation screening or tests that	t Number of tests in denominator completed within 8	Number of Mutation screening or tests that require Southern blot	Quarterly	Quarterly	Laboratory reports	5		н	figher is better	Numerator / Denominator		A normal Fragile X result would be reported within 2 (or 4) weeks (specific	Include all tests in the lab, not just those which are	
	recover from episodes of ill health or following injury		require Southern blot analysis completed within 8 weeks	weeks (40 working days)	analysis								x 100		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	requested by clinical genetics.	
															time would apply for the entire test. This also applies to myotonic dystrophy and Huntington disease.		
GEN04by	Domain 3: Helping people to recover from episodes of ill health	Laboratory reporting times	Proportion of next generation sequencing panels of >10 genes completed within 16	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	2		н	ligher 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	or following injury Domain 5: Treating and caring for	Educational sessions provided by	weeks Rate of educational sessions provided by	Number of educational sessions provided by clinical		Quarterly	Quarterly	Hospital records			т	There is a need to assess			An educational session is a formal post graduate education session lasting an an		
	people in a safe environment and protecting them from avoidable	clinical genetics to other specialties to support genetics in	clinical genetics to other specialties	genetics to other specialties	(excluding trainees)							everage number of educational sessions in			hour (excludes undergraduate teaching)		
	harm	mainstream medicine									e	ime period that would be expected as good practice					
											-	n order to set a range for nterpretation guidance.					
GEN06	Domain 4: Ensuring that people	Poor patient experience	Pate of written complaints about the renetic	Number of written complaints about the genetics	Total number of patient contacts (appointments including	Quarterly	Quarterly	Hospital records				ower is better	Numerator / Denominator				
	have a positive experience of care	roor patient experience	department	department received during period	telephone contacts that replace a face-to-face appointment) in period	Quarterly	Quarterry	nospital records			Ĩ		x 100				
GEN07	Domain 4: Ensuring that people have a positive experience of care	Good patient experience	Rate of letters/emails from patients, carers o non-genetics consultants registering thanks t	r Number of letters/emails from patients, carers or non- o genetics clinicians registering thanks to the genetics	Total number of patient contacts (appointments including telephone contacts that replace a face-to-face appointment) in	Quarterly	Quarterly	Hospital records			н	figher is better	Numerator / Denominator x 100				
			the genetics department	department received during period	period												
GEN08	Domain 4: Ensuring that people have a positive experience of care	pre-natal (PN) genetic test results	within 5 working days after the clinic recieves	diagnosis during the period, the number who received	Number of patients seen in clinical genetics who had prenatal diagnosis during period	Quarterly	Quarterly	Hospital clinical records			н	ligher 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		
		where the patient is seen in the clinical genetics department	the laboratory report for PN genetic test results	their prenatal genetic test result within 5 working days of the clinic receiving the laboratory report.											face-to-face and more likely for PN test results this is done over the phone by a consultant geneticist or genetic coursellor. 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	in the Data Dictionary	Proportion of appointments that are not attended	Number of DNAs within period	clinical genetics (311 treatment function). This can be taken as	Quarterly	Quarterly	Hospital clinical records			L	ower is better	Numerator / Denominator x 100				
		(http://www.datadictionary.nhs. uk/data_dictionary/attributes/a/			the total number recorded on PAS including DNAs												
		ari/attended_or_did_not_attend _de.asp?shownav=1)															
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			L	ower is better	Numerator / Denominator x 100		Trends need to be monitored		
GEN11a	have a positive experience of care Domain 4: Ensuring that people		Rate of patients co-counselled by a penetic	The number of clinic appointments where a network	Total number of clinical genetics appointments of all types during	Quarterly	Quarterly	records Hospital clinical				IBC III		It is recognised that	Ideally this would be an average of rates measured per patient but it's recognised		
	have a positive experience of care		counsellor and doctor during period	counsellor co-counsels with a doctor (excluding MDC co	 period (appointments including telephone contacts that replace a face-to-face appointment) 	,	and only	records				-		hospital IT systems may not record	locally 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.		
														coursellor non- consultant			
														appointments therefore this			
														measure is intended to			
														promote better recording of these			
C	Demain & Ferry	Definete et	Date of equipterants	The sumber of indexed as a set of the set of	Total number of clinical genetics appointments of all types during	Quantanta.	0	Managhad 1911-1						appointments.			
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	appointments (excluding planned pre-clinic	Total number of clinical genetics appointments of all types during period (appointments including telephone contacts that replace a face-to-face appointment)	Quarteny	Quarterly	Hospital clinical records			T	DC		It is recognised that hospital IT systems may not record	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.		
		COM GENU	procession and a second s											may not record counsellor non- consultant			
														appointments therefore this			
														measure is intended to			
														promote better recording of these			
				-			-							appointments.			
GEN11c	Domain 4: Ensuring that people have a positive experience of care	Patient appointments held in preparation for a doctor's	Rate of genetic counsellor clinic appointments held in preparation for a	held in preparation for a subsequent doctor's	Total number of clinical genetics appointments of all types during period (appointments including telephone contacts that replace a fore to fore appointment).	Quarterly	Quarterly	Hospital clinical records			т	BC		It is recognised that hospital IT systems	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.		
		appointment	doctor's appointment	appointment	face-to-face appointment)									may not record coursellor non-			
														consultant appointments therefore this			
														therefore this measure is intended to			
				1	1									promote better recording of these			
							1	1	1					appointments.			
<u>GEN12a</u>	Domain 5: Treating and caring for people in a safe environment and	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.		
GEN12a	people in a safe environment and protecting them from avoidable harm	regarding patient care	involving patient care	care				system							depending on the incident.		
GEN12a GEN12b	people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and		involving patient care	care		Quarterly Quarterly		SUI reporting system SUI reporting system									
	people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm	regarding patient care Serious untoward Incidents regarding lab tests	involving patient care Rate of serious Untoward Incidents involving lab tests	care Number of serious Untoward Incidents involving lab tests	Number of lab tests during period	Quarterly	Quarterly	system SUI reporting system							depending on the incident. Suggest we review against patient numbers seen / number of lab reports - depending on the incident.		
GEN12a GEN12b GEN13	people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and	regarding patient care Serious untoward Incidents	involving patient care Rate of serious Untoward Incidents involving	care	Number of lab tests during period			system SUI reporting system							depending on the incident. Suggest we review against patient numbers seen / number of lab reports - depending on the incident. It is mandatory that all labs participate in at least one EQA science. The EQA schemes are tet specific by condition on therefore we would expert the specific		
	people in a safe environment and protecting them from avoidable harm. Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm. Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm.	regarding patient care Serious untoward incidents regarding lab tests External Quality Assurance	involving patient care Rate of serious Untoward Incidents involving lab tests Episodes of poor performance in EQA schemes the laboratory participates in	care Number of serious Untoward Incidents involving lab tests Episodes of accor performance notified to the laboration by UINEQAS, EMQN or another EQA service provider	Number of lab tests during period Number of IQA schemes participated in	Quarterly Annual (FY)	Quarterly Annual (FY)	system SUI reporting system EQA							depending on the incident. Suggest we review against patient numbers seen / number of lab reports - depending on the incident. It is mandratory that all labs participate in at least one EQA scheme. The EQA cohemes are test pacific by condition and herefore we would be the sportifi- schemes to vary by bia sa different labs across the country provide different tests of afferent condition.		
GEN13	people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and	regarding patient care Serious untoward Incidents regarding lab tests	involving patient care Rate of serious Untoward Incidents involving lab tests Episodes of poor performance in EQA schemes the laboratory participates in	are Insuface of serious Unitoward incidents involving lab tests Episodes of poor performance notified to the laboratory UNIXIGAX_BURGY or another EGA series provider Number of audits (from the agreed list of audits) that	Number of lab tests during period Number of EQA schemes participated in	Quarterly	Quarterly Annual (FY)	system SUI reporting system							depending on the incident. Suggest we review against patient numbers seen / number of lab reports - depending on the incident. It is manufatory that all labs participate in at least one EQA scheme. The EQA schemes are test specific by condition and therefore we would expert the specific schemes to any by this ad different lass on the county provide different tests		
GEN13	people in a safe environment and protecting them from an avoidable harm Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable Domain 5: Treating and caring for portecting at the monoument to harm the monoument and harm the monoument and the harm and the monoument and the monoument and the monoument the monoument and the monoument and the monoument and the harm and the monoument and the monoument and the monoument and the harm and the monoument and the monoument and the monoument and the harm and the monoument and the monoument and the monoument and the monoument and the harm and the monoument and th	regarding patient cure Serious untoward incidents regarding lab tests External Quality Assurance Activity audits (laboratory) Non-Adherence to UKGTN	Involving patient care Rate of serious Untoward incidents involving lab tests Epocodes of poor performance in EQA schemes the laboratory participated in by the generics laboratory in number and type of audia series to be agreed) Proportion of audits participated in by the generics laboratory the number and type of audia series to be agreed)	care Number of serious Unitoward incidents involving lab tests Episodes of poor performance notified to the laboratory by UNNEQAS, EMON or another EQA service provider Number of audits (from the agreed list of audits) that pencies laboratory participates in Number of tests requested by clinical genetics from	Number of lab tests during period Number of EQA schemes participated in Number of audits that the genetics laboratories should be participating in	Quarterly Annual (FY)	Quarterly Annual (FY)	system SUI reporting system EQA Dependent on specific audits				ower is better			depending on the incident. Suggest we review against patient numbers seen / number of lab reports - depending on the incident. It is mundatory that all labs participate in at least one EQA scheme. The EQA schemes are test pacific by condition on therefore we would expert the specific schemes to vary by bla sa different labs across the county provide different tests for different conditions. As per Cinculal Wolecular General Sacktry/Association for Clinical Optigenetics and UKDT having Citeria are available from the UKDTN website. The 3 tests for	to include workforce audit	
GEN13	people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm Domain 5: Treating and caring for people in a safe environment and protecting them from avoidable harm	regarding patient cure Serious unitoward incidents regarding lab tests Estemal Quality Assurance Activity audits (laboratory)	Involving patient care Rate of serious Untoward incidents involving lab tests Episodes of poor performance in EQA schemes the balocatory participates in Proportion of saddts participate in by the generics labolatory for mumber and type of audits need to be agreed]	are Number of serious Untoward incidents involving lab tests Episoden of goor performance notified to the laboratory by UREQAS, EMQL or another EDA revice provider Number of audits (from the agreed list of audits) that genetics laboratory participates in	Number of lab tests during period Number of EQA schemes participated in Number of audits that the genetics laboratories should be participating in	Quarterly Annual (PY) Annual (PY)	Quarterly Annual (FY) Annual (FY)	system SUI reporting system EQA Dependent on specific audits agreed				ower is better			depending on the incident. Suggest we review against patient numbers seen / number of lab reports - depending on the incident. It is mediatory that all labs participate in at least one EQA scheme. The EQA schemes are then specific by condition and therefore we would report the specific resolutions with the specific by condition and therefore we would report the specific of different conditions. As per Clinical Molecular Generics Society/Association for Clinical Cytogenetics and UKGTN audits requested	to include workforce audit	