

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
RECOMMENDATIONS TO BE CONSIDERED FOR FUNDING											
#185	Lipoprotein Lipase Deficiency (LPL)	Aberdeen RGC	£525	£140	300	900	Consultant Metabolic Medicine/Chemical Pathology Consultant Clinical Geneticist Consultant Metabolic Paediatrician	E01: Medical Genetics (I=0, F=600) E06: Metabolic disorders (I=300, F=250) E03: Paediatric Medicine (I=50, F=50)	Costs: £264,750	Costs: £84,000	E06 Costs: £153,928 E03 Costs: £26,822
#186	Chromosome Breakage 89 Gene Panel Test	Bristol RGC	£900	£190	50	100	Consultant Clinical Geneticist Consultant Haematologist Consultant in Bone Marrow Transplantation	E01 Medical Genetics (I=30, F=60) E04 Paediatric cancer services (I=20, F=40)	Costs: £41390	Costs: 24,834	Costs: £16,556
#187 (RARE)	GATA2 deficiency	Bristol RGC	£459	£190	10	30	Consultant Haematologist Consultant Clinical Geneticist Consultant Oncologist	E01. Medical Genetics (I=2, F=6) B04. Blood & Marrow Transplantation (I=6, F=24) B03. Specialised cancer (I=2, F=0)	Savings: £5,798	Savings: £1159.60	B04 savings £2338.80 B03 savings £2299.60
#188	Very Early Onset Inflammatory Bowel Disease 40 Gene Panel Test	London North East RGC GOSH	£1,250	£130 (1 amplicon) £185 (2 amplicons)	40	10	Consultant Clinical Geneticist Consultant Paediatric Gastroenterologist	E01 Medical Genetics (I=0, F=6) E03 Paediatric medicine (I=40, F=4)	Savings: £50,920	Costs: £888	Savings: £51,808
#190	Syndromic & Non Syndromic Hearing Loss 95 Gene Panel Test	London North East RGC GOSH	£1,500	£130/£185	50	20	Consultant Clinical Geneticist Consultant Audiologist Consultant Paediatrician	E01 Medical Genetics (I=30, F=20) CCG Audiology (I=20, F=0)	Savings: £6265	Savings: £2279	Savings: £3986

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
#193	HYPOCALCAEMIC HYPERCALCAEMIA, FAMILIAL, TYPE III; HHC3	Oxford RGC	£153	£153	100	40	Consultant Endocrinologist Consultant Clinical Geneticist Consultant Biochemist	E01: Medical Genetics (I=50, F=25) A03: Specialised Endocrinology (I=50, F=15)	Costs: £21420	Costs: £11475	Costs: £9945
#194 (RARE)	Rhabdoid Tumour Predisposition Syndrome 1	London North East RGC GOSH	£460	£130	10	5	Consultant Clinical Geneticist Consultant Paediatric Oncologist	E01: Medical Genetics (I=5, F=5) CCG Paediatric oncology (I=5, F=0)	Costs: £3250	Costs: £1950	Costs: £1300
#201 (multi gene) (Evaluate with #213 - Guy's Panel Test)	Urea cycle disorders (UCDs)	Sheffield RGC	£600	£105	48	80	Consultant in Metabolic Disease (Paediatric/Adult) Consultant Paediatrician Consultant Neonatologist Consultant Hepatologist Consultant Neurologist Consultant Clinical Geneticist	E01 Medical Genetics (I=5, F= 35) E06 Metabolic Disorders (I=35, F=45) E03 Paediatric Medicine (I=5, F=0) E09 Paediatric neuroscience (I=3, F=0)	Costs: £1826	Costs: £2990	E06 savings: £69 E03 savings: £685 E09 savings: £410
#202	Monogenic Diabetes 14 Gene Panel Test	Exeter RGC	£650 or if test following sanger sequencing then £300	£100	200	49	Consultant Clinical Geneticists Consultant Paediatricians Consultants in Endocrinology & Diabetes Consultants in General Medicine Consultant Biochemists Consultant Nephrologists GP Principle in liaison with genetic diabetic nurses	E01 Medical Genetics (I=20, F=20) CCG Endocrinology & Diabetes (I=130, F=15) CCG Paediatrics (I=20, F=5) CCG Genetic Diabetes Nurses referring through General Practice (I=5, F=5) CCG Clinical Biochemistry (I=10, F=2) CCG General Medicine (I=10, F=2) CCG Nephrology (I=5, F=0)	Cost neutral	Cost neutral	Cost neutral
#203	Neonatal Diabetes 29 22 Gene Panel Test	Exeter RGC	£650	£100	20	40	Consultant Endocrinologists Consultant Paediatricians Consultant Clinical Geneticists	E01 Medical Genetics (I=3, F=6) CCG Endocrinology & Diabetes (I=12, F=24) CCG Paediatrics (I=5, F=10)	Costs £5,335	Costs £800	CCG Endocrinology & Diabetes £3,201 CCG Paediatrics £1,334

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
#204	Hereditary Cancer and Fanconi Anaemia TruSIGHT 94 Gene Panel Test	Sheffield RGC	Hereditary Cancer panels: £800 Fanconi anaemia panel: £1000	£155	BRCA1/2: 8400 Colorectal cancer: 2,400 Pheochromocytoma/paraganglioma panel: 415 Renal cancer panel local activity 5 Fanconi anaemia panel: 6	BRCA1/2: 3400 Colorectal cancer: 845 Pheochromocytoma/paraganglioma panel: 300 Renal cancer panel local approx 12 Fanconi anaemia panel: 4	Consultant Clinical Geneticist Consultant Clinical Nephrologist Consultant Clinical Oncologist Consultant Clinical Endocrinologist Consultant Clinical Haematologist	E01 Medical Genetics B03 Specialised Cancer B14 Specialised Urology A03 Specialised Endocrinology CCG	Cost neutral	Cost neutral	Cost neutral
#207	Primary Ciliary Dyskinesia 18 Gene Panel Test	Leeds RGC	£1,250	£220	75	40	Consultant Clinical Geneticist Consultant Paediatrician Consultant in Respiratory Medicine	E01 Medical Genetics (I=0, F=40) E03 Paediatric Medicine (I=74, F=0) A14 Specialised Respiratory (I=1, F=0)	Costs: £18,050	Costs £6800	E03 costs £11,100 A14 costs £150
#209 (RARE)	Nonsyndromic Holoprosencephaly	Exeter RGC	£450	£105	5	10	Consultant Clinical Geneticist	E01: Medical Genetics	Costs: £3300	Costs: £3300	n/a
#210 (RARE)	Congenital fibrosis of the extraocular muscles 2 (CFEOM2)	Exeter RGC	£105	£100	5	10	Consultant Neurologist Consultant Ophthalmologist Consultant Clinical Geneticist	E01. Medical Genetics (I=1, F=6) D04. Neurosciences (I=2, F=2) D12. Specialised ophthalmology services (I=2, F=2)	Costs: £1225	Costs: £645	D04 costs: £290 D12 costs: £290
#211	Primary pigmented nodular adrenocortical disease (PPNAD)	Exeter RGC	£1,100	£105	5	5	Consultant Clinical Endocrinologist Consultant Clinical Geneticist	E01: Medical Genetics (I=2, F=5) A03: Specialised Endocrinology (I=3, F=0)	Costs: £4425	Costs: £2085	Costs: £2340

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
#213 (Evaluate with #201 - Sheffield multi gene)	Urea Cycle Defects (UCD) 9 Gene Panel Test	London South East RGC GSTT	£850	£160	47	80	Consultant Clinical Geneticist Consultant in Metabolic Medicine (Paediatric/Adult) Consultant Hepatologist Consultant Paediatrician Consultant Neonatologist Consultant Neurologist	E01 Medical Genetics (I=5, F= 35) E06 Metabolic Disorders (I=35, F=45) E03 Paediatric Medicine (I=5, F=0) E09 Paediatric neuroscience (I=5, F=0)	Costs: £7300	Costs: £5050	E06 costs: £3350 E03 savings: £550 E09 savings: £550
#214	Alport Syndrome 5 Gene Panel Test	London South East RGC GSTT	£750	£160	134	225	Consultant Clinical Geneticist Consultant Nephrologist	E01 Medical Genetics A06 Renal Dialysis	Savings £176,118	Savings £86,059	Savings: £90,059
#215	Methylmalonic acidemia (MMA) 11 Gene Panel Test	London South East RGC GSTT	£850	£160	30	60	Consultant in Metabolic Medicine Consultant Clinical Geneticist Consultant in Hepatology	E01 Medical Genetics (I=0, F=60) E06 Metabolic Disorders (I=30, F=0)	Savings £11,576	Costs £9600	Savings £21176
#216	Inherited Ataxias 57 Gene Panel Test	Oxford RGC	£1,020	£170	100	10	Consultant Clinical Geneticists Consultant Neurologists Consultant Paediatric Neurologists	E01 Medical Genetics (I=60, F=10) D04 Neurosciences (I=25, F=0) E09 Paediatric neurosciences (I=15, F=0)	Savings: £294,300	Savings: £175,900	D04 savings: £74000 E09 savings: £44,400
#217	KBG syndrome (short stature, macrodontia, characteristic facies, skeletal abnormalities and developmental delay	Bristol RGC	£550 TBC	£190	10	20	Consultant Clinical Geneticist	E01: Medical Genetics	Savings: £3562	savings: £3562	n/a
#221 (RARE)	CHILD syndrome	Exeter RGC	£350	£100	5	10	Consultant Clinical Geneticist	E01: Medical Genetics	Costs: £2750	Costs: £2750	n/a
#222 (RARE)	Familial hyperphosphatemic tumoral calcinosis (HFTC)	Exeter RGC	£350	£100	5	10	Consultant Clinical Geneticist	E01: Medical Genetics	Costs: £4000	Costs: £4000	n/a

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
#223	Osteopetrosis 21 Gene Panel test	Bristol RGC	£600	£190	10	20	Consultant paediatricians specialising in bone marrow transplantation, haematology, metabolic disease, or orthopaedics Consultant Clinical Geneticists Adult Orthopaedic Consultants (ADO only)	E01 Medical Genetics (I=5, F=20) CCG Paediatrics (I=3, F=0) CCG Orthopaedics (I=2, F=0)	Savings £56225	Savings £26,687	CCG Paediatrics savings £17723 CCG Orthopaedics savings £11815
#224 (RARE)	Galactosialidosis	London South East RGC GSTT	£450	£160	5	20	Consultant in Inherited Metabolic Disease Consultant Clinical Geneticist	E01 Medical Genetics (I=0, F=15) E06 Metabolic disorders (I=5, F=5)	Costs £5450	Costs £2400	Costs £3050
#225	GM1 gangliosidosis and mucopolysaccharidosis type IVB	London South East RGC GSTT	£450	£160	20	60	Consultant in Inherited Metabolic Disease Consultant Clinical Geneticist	E01 Medical Genetics (I=5, F=45) E06 Metabolic disorders (I=15, F=15)	Costs £18600	Costs £9450	Costs £9150
#227	Lysosomal acid lipase deficiency	London South East RGC GSTT	£375	£160	10	20	Consultant Clinical Geneticist Consultant Metabolic Disease Specialist Consultant Chemical Pathologist Consultant Lipidologist	E01 Medical Genetics (I=0, F=12) E06 Metabolic disorders (I=4, F=8) CCG Chemical Pathology (I=2, F=2) CCG Lipidology (I=4, F=3)	Costs £7750	Costs £1920	E06 Metabolic disorders costs £2780 CCG Chemical pathology costs £1070 CCG Lipidology costs £1980
#228 (RARE)	Neuraminidase deficiency	London South East RGC GSTT	£375	£160	5	20	Consultant in Inherited Metabolic Disease Consultant Clinical Geneticist	E01. Medical Genetics (I=0, F=12) E06. Metabolic disorders (I=5, F=8)	Costs £5075	Costs £1920	Costs £3155
#229	Pyruvate carboxylase deficiency	London South East RGC GSTT	£450	£160	5	20	Consultant in Inherited Metabolic Disease Consultant Clinical Geneticist	E01 Medical Genetics (I=1, F=10) E06 Metabolic Disorders (I=4, F=10)	Savings £1050	Costs £750	Savings £1800

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
#234	Paediatric Cardiomyopathy 73 Gene Panel Test	Bristol RGC	£900	£190	100	300	Consultant Clinical Geneticist Consultant Paediatric Cardiologist in liaison with Clinical Genetics Consultant Pathologist in liaison with Clinical Genetics Paediatric Neuromuscular Consultant in liaison with Clinical Genetics Paediatric Metabolic Consultant in liaison with Clinical Genetics	E01 Medical Genetics E05 Congenital Heart Services E09 Paediatric neurosciences E06 Metabolic disorders Medical Pathology	Cost neutral	Cost neutral	Cost neutral
#236	Neurogenetic Disorders 95 Gene Panel Test	Sheffield RGC	Ataxia and Hereditary Spastic Paraplegia subpanels £1,000 Dystonia and Parkinsonism subpanel £850 Amyotrophic Lateral Sclerosis / Frontotemporal Dementia and Hemiplegic Migraine subpanels £800	£105	750	376	Consultant Clinical Geneticists Consultant Neurologists Consultant Paediatric Neurologists	E01 Medical Genetics (I=55, F=320) D04 Neurosciences (I=525, F=38) E09 Paediatric neurosciences (I=170, F=18)	Savings £67456	Costs £19306	D04 savings £65897 E09 savings £20865
#239	Congenital Anaemia 33 Gene Panel Test	Oxford Haemoglobinopathy	£300	£200	100	60	Consultant Paediatric Haematologist Consultant Haematologist Consultant Clinical Geneticist	E01, Medical Genetics (I=5, F=3) E03 Paediatric Medicine (I=80, F=48) CCG Haematology (I=15, F=9)	Savings £25000	Savings £1250	E03 savings £20,000 CCG Haematology savings £3750
#240	Iron Regulatory 16 Gene Panel Test	Oxford Haemoglobinopathy	£300	£200	175	10	Consultant Haematologist Consultant Gastroenterologist Consultant Hepatologist Consultant Clinical Geneticist Consultant Endocrinologist Consultant Rheumatologist	E01 Medical Genetics (I=3, F=10) CCG Gastroenterology (I=42, F=0) CCG Hepatology (I=21, F=0) CCG Haematology (I=102, F=0) CCG Rheumatology (I=2, F=0) A03 Specialised endocrinology (I=5, F=0)	Savings £97,800	Savings £1480	CCG Gastroenterology savings £23,520 CCG Hepatology savings £11760 CCG Haematology savings £57,120 CCG Rheumatology savings £1120 A03 Specialised endocrinology savings £2800

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
#242 (RARE)	HYPEREKPLEXIA, HEREDITARY 1; HKPX1 HYPEREKPLEXIA 3; HKPX3 HYPEREKPLEXIA 2; HKPX2	Cardiff RGC	£620	£160	15	20	Consultant Clinical geneticist Consultant Neurologist Consultant Paediatric Neurologist	E01. Medical Genetics (I=5, F=20) E09. Paediatric Neurosciences (I=1, F=0) CCG Neurology (I=9, F=0)	Costs £12500	Costs £6300	E09 costs £620 CCG costs £5580
#250	Early Infantile Epileptic Encephalopathy (EIEE) 45 Gene Panel Test	London North East RGC GOSH	£1,250	£130	300	75	Consultant Clinical Geneticist Consultant Paediatric Neurologist	E01. Medical Genetics (I=200, F=50) CCG Paediatric Neurology (I=100, F=25)	Savings £303,980	Savings £202,653	Savings £101,327
#251	Cancer 83 Gene Panel Test	Leeds RGC	£860	£170	BRCA1/2: 8400 Colorectal cancer: 2,400 Phaeochromocytoma/paraganglioma panel: 415 Renal cancer panel local activity 5 Fanconi anaemia panel: 6	BRCA1/2: 3400 Colorectal cancer: 845 Phaeochromocytoma/paraganglioma panel: 300 Renal cancer panel local approx 12 Fanconi anaemia panel: 4	Consultant Clinical Geneticist Consultant Clinical Nephrologist Consultant Clinical Oncologist Consultant Clinical Endocrinologist Consultant Clinical Haematologist	E01 Medical Genetics B03 Specialised Cancer B14 Specialised Urology A03 Specialised Endocrinology CCG	Cost neutral	Cost neutral	Cost neutral
#253	Cardiac 67 Gene Panel Test	Manchester RGC	1. Aortic aneurysm (AA) £750 2. Arrhythmogenic right ventricular cardiomyopathy (ARVC) £700 3. Brugada Syndrome £700 4. Catecholaminergic polymorphic ventricular tachycardia (CPVT) £700 5. Dilated cardiomyopathy (DCM) £800 6. Hypertrophic cardiomyopathy (HCM) £800 7. Long QT syndrome (LQT) £750 8. Cardiomyopathies (CM) £900 9. Arrhythmia/cardiac arrest (BCL) £800 10. Molecular Autopsy (MolAut) £1100	£138	750	750	Consultant Clinical Geneticist Consultant Cardiologist	E01: Medical Genetics (I=735, F=750) CCG Cardiology (I=15, F=0)	Cost neutral	Cost neutral	Cost neutral
GENE DOSSIERS SUBMITTED AND RECOMMENDED FOR HIGHLY SPECIALISED SERVICES (PREVIOUSLY NSCT) FUNDING											

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
#189	Bardet-Biedl Syndrome 13 Gene Panel Test	London North East RGC GOSH	£1,250 (if not covered by HSS funding – i.e. patient NOT resident in England or Scotland)	£130 or £185 (if not covered by HSS funding – i.e. non-parental carrier tests)	20	15	Consultant Clinical Geneticist Consultant Ophthalmologist Consultant Nephrologist	HSS Genetics (I=10, F=15) HSS Ophthalmology (I=5, F=0) HSS Nephrology (I=5, I=0)	Savings £11,800	n/a	HSS savings £11800
#195 (PART) #233 assigned also as duplicate	Connective Tissue Disorders 67 Gene Panel Test	Sheffield RGC	Average across the sub panels of £925	£155	965	345	Consultant Clinical Geneticist Consultant Neurologist Consultant Paediatrician Consultant Nephrologist Consultant Ophthalmologist Consultant Orthopaedist Consultant Paediatric Neurologist Consultant Neonatologist Consultant in Stroke Medicine Consultant Dermatologist Consultant Cardiologist Consultant in Respiratory Medicine Consultant in Bone Metabolism/Endocrinologist	E01 Medical Genetics (I=965, F=345) HSS Clinical Genetics (I=150, F=0) D04 Neurosciences (I=28) E03 Paediatric Medicine (I=30) E09 Paediatric neurosciences (I=7) E08 Neonatal Critical Care (I=5) B14 Specialised urology (I=10) A12 Specialised Dermatology (I=77) A14 Specialised Respiratory (I=2) A09 Complex Invasive Cardiology (I=6) D12 Specialised Ophthalmology (I=10) D10 Specialised Orthopaedic Services (I=4) A03 Specialised Endocrinology (I=5) HSS Metabolic bone/endocrinology (I=15) CCG Consultants in Stroke Medicine (I=2)	Savings £1,117,757	Savings £711,195	HSS Clinical Genetics savings £173,745 D04 Neurosciences savings £32,432 E03 Paediatric Medicine savings £34,749 E09 Paediatric neurosciences savings £8108 E08 Neonatal Critical Care savings £5791 B14 Specialised urology savings £11583 A12 Specialised Dermatology savings £89,189 A14 Specialised Respiratory savings £2317 A09 Complex Invasive Cardiology savings £6950 D12 Specialised Ophthalmology savings £11583 D10 Specialised Orthopaedic Services savings £4633 A03 Specialised Endocrinology savings £5791 HSS Metabolic bone/endocrinology savings £17374 CCG Consultants in Stroke Medicine savings £2317

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
#198 (PART)	GSD 32 Gene Panel Test	Sheffield RGC	800	105	75	30	Consultant Paediatrician Consultant Neurologist Consultant in Metabolic Medicine Consultant Clinical Geneticist Consultant Hepatologist	E01. Medical Genetics (I=10, F=17) E03. Paediatric medicine (I=15, F=3) E06. Metabolic disorders (I=30, F=10) D04. Neurosciences (I=15, F=0) A02. Hepatobiliary and pancreas (I=5, F=0) Proportion of above will be for HSS where test referred from the National Diagnostic and Management Centre for McArdle Disease related disorders	Cost neutral	Cost neutral	Cost neutral
#199 (RARE)	CEROID LIPOFUSCINOSIS, NEURONAL, 7; CLN7	London North East RGC GOSH	£460 (usually requested alongside CLN5, CLN6, and CLN8 – total cost £675)	£130 / £185 for 1 or 2 amplicons, respectively	10	11	Consultant Neurologist Consultant Clinical Geneticist Consultant Metabolic Paediatrician	HSS Lysosomal Storage Disorders Service	Costs £3980	na/	HSS costs £3980
#205	CRANIOSYNOSTOSIS AND DENTAL ANOMALIES; CRSDA	Oxford RGC	£510 (HSS)	£153	20	2	Consultant Clinical Geneticist	HSS	Costs: £9026	HSS costs £9026	n/a
#206 (PART)	Primary Immune Deficiencies 72 Gene Panel Test (part HSS)	London North East RGC GOSH	£1,250	£130 (single mutation); £185 (two mutations)	40	20	Consultant clinical geneticist Consultant immunologist	E01. Medical Genetics (I=0, F=20) B09. Specialised immunology & allergy services (I=20, F=0) HSS (I=20, F=0)	Savings £34,410	Costs £1850	B09 savings £18,130 HSS savings £18,130
#248 (RARE) (PART)	Short Syndrome	Cambridge RGC	£415	£145	10	17	Consultant Endocrinologist Consultant Paediatrician Consultant Clinical Geneticist Consultant for NHS National Severe Insulin Resistance Service	E01 Medical Genetics (I=6, F=8) CCG Endocrinology (I=1, F=2) CCG Paediatrics (I=1, F=2) HSS National Severe Insulin Resistance Service (I=2, F=5)	Savings £2135	Savings £1600	CCG endocrinology savings £170 CCG Paediatrics savings £170 HSS savings £195
NON-INVASIVE PRENATAL DIAGNOSIS (NIPD) - FOR NEW CONDITIONS											

Gene Dossiers Evaluations for 2015/16 Commissioning Year

Evaluation number as assigned during the process	Test Name	Laboratory	Cost per test for index cases	Cost per test for family members	Expected annual referrals for index cases	Expected annual referrals for family members	Types of referrers on testing criteria	Split of activity based on CRG	Estimate of total UK annual costs/savings based on expected numbers	Estimate of UK annual costs/savings from clinical genetics	Estimate of UK annual costs/savings from mainstream specialty outside of clinical genetics
#191 (NIPD)	Cystic Fibrosis; CF	London North East RGC GOSH	£550	N/A	5	N/A	Consultant Clinical Geneticist Consultant in Fetal Medicine	E01: Medical Genetics (I=2, F=0) E12: Fetal Medicine (I=3, F=0)	Costs: £385	Costs: £154	Costs: £231
#200 (NIPD)	Apert syndrome- using cell free fetal DNA (cffDNA) for non-invasive prenatal diagnosis (NIPD) for 2 common FGFR2 mutations.	London North East RGC GOSH	£550	TBC	4	TBC	Consultant Clinical Geneticist Consultant in Fetal Medicine	E01: Medical Genetics (I=4, F=0) E12: Fetal Medicine (I=4, F=0)	Costs: £2508	Costs: £1254	Costs: £1254