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| **WGS Phase 1 Rare Disease Clinical Indications - March 2021** | | |
| 21 clinical indications for WGS | | |
| **Clinical indication ID** | **Clinical Indication** | **GMS PanelApp link** |
| R89 | Ultra-rare and atypical monogenic disorders | Clinical Genetics Only - Relevant panel(s) can be chosen from <https://nhsgms-panelapp.genomicsengland.co.uk/> |
| R27 | Congenital malformation and dysmorphism syndromes | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/486/v14.43> |
| R100 | Rare syndromic craniosynostosis or isolated multisuture synostosis | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/168/v2.2> |
| R104 | Skeletal dysplasia | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/309/v2.2> |
| R109 | Childhood onset leukodystrophy | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/496/v6.20> |
| R143 | Neonatal diabetes | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/293/v2.2> |
| R193 | Cystic renal disease | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/487/v3.19> |
| R29 | Intellectual disability – microarray, fragile X and sequencing | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/285/v3.2> |
| R381 | Other rare neuromuscular disorders | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/465/v5.43> |
| R54 | Hereditary ataxia with onset in adulthood | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/466/v2.13> |
| R55 | Hereditary ataxia with onset in childhood | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/488/v6.51> |
| R59 | Early onset or syndromic epilepsy | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/402/v2.2> |
| R61 | Childhood onset hereditary spastic paraplegia | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/568/v2.18> |
| R69 | Hypotonic infant with a likely central cause | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/490/v9.41> |
| R83 | Arthrogryposis | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/258/v3.2> |
| R84 | Cerebellar anomalies | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/488/v6.51> |
| R85 | Holoprosencephaly - NOT chromosomal | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/78/v2.3> |
| R86 | Hydrocephalus | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/179/v2.3> |
| R87 | Cerebral malformation | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/491/v5.17> |
| R88 | Severe microcephaly | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/162/v2.2> |
| R98 | Likely inborn error of metabolism | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/846/v1.36> |
| **WGS Phase 2 Rare Disease Clinical Indications – cohort 1 (4th October 2021)** | | |
| 15 further clinical indications for WGS | | |
| R56 | Adult onset dystonia, chorea or related movement disorder v1.2 (R56.1) | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/540/v1.121> |
| R57 | Childhood onset dystonia, chorea or related movement disorder v1.2 (R57.1) | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/847/v1.137> |
| R58 | Adult onset neurodegenerative disorder v2.2 (R58.1) | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/474/v2.178> |
| R60 | Adult onset hereditary spastic paraplegia v1.3 (R60.1) | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/567/v1.27> |
| R62 | Adult onset leukodystrophy v1.2 (R62.1) | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/579/v1.25> |
| R78 | Hereditary neuropathy or pain disorder v1.2 (R78.1) | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/846/v1.36> |
| **WGS Phase 2 Rare Disease Clinical Indications – cohort 2 (25th October 2021)** | | |
| R135 | Paediatric or syndromic cardiomyopathy v1.4 (R135.1) | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/749/v1.4> |
| R257 | Unexplained paediatric onset end-stage renal disease | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/678/v1.2> |
| **WGS Phase 2 Rare Disease Clinical Indications – cohort 3 (22nd November 2021)** | | |
| R15 | Primary immunodeficiency | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/398/v2.1> |
| **WGS Phase 2 Rare Disease Clinical Indications – cohort 4 (4th January 2022)** | | |
| R31 | Bilateral congenital or childhood onset cataracts | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/230/v2.76> |
| R32 | Retinal disorders | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/307/v2.195> |
| R33 | Possible X-linked retinitis pigmentosa | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/307/v2.195> |
| R34 | Sorsby retinal dystrophy | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/307/v2.195> |
| R35 | Doyne retinal dystrophy | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/307/v2.195> |
| R36 | Structural eye disease | <https://nhsgms-panelapp.genomicsengland.co.uk/panels/509/v1.3> |