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| **WGS Rare Disease Clinical Indications - March 2021** |
| 21 clinical indications for WGS |
| **Clinical indication ID** | **Clinical Indication** | **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 | [R27 Paediatric disorders](https://panelapp.genomicsengland.co.uk/panels/486/) |
| R100 | Rare syndromic craniosynostosis or isolated multisuture synostosis | [R100 Craniosynostosis](https://panelapp.genomicsengland.co.uk/panels/168/) |
| R104 | Skeletal dysplasia | [R104 Skeletal dysplasia](https://panelapp.genomicsengland.co.uk/panels/309/) |
| R109 | Childhood onset leukodystrophy | [R109 White matter disorders - childhood onset](https://panelapp.genomicsengland.co.uk/panels/496/) |
| R143 | Neonatal diabetes | [R143 Diabetes - neonatal onset](https://panelapp.genomicsengland.co.uk/panels/293/) |
| R193 | Cystic renal disease | [R193 Cystic renal disease](https://panelapp.genomicsengland.co.uk/panels/487/) |
| R29 | Intellectual disability – microarray, fragile X and sequencing | [R29 Intellectual disability](https://panelapp.genomicsengland.co.uk/panels/285/) |
| R381 | Other rare neuromuscular disorders | [R381 Neuromuscular disorders (Super Panel)](https://panelapp.genomicsengland.co.uk/panels/465/) |
| R54 | Hereditary ataxia with onset in adulthood | [R54 Hereditary ataxia - adult onset](https://panelapp.genomicsengland.co.uk/panels/466/) |
| R55 | Hereditary ataxia with onset in childhood | [R55 Hereditary ataxia and cerebellar anomalies - childhood onset](https://panelapp.genomicsengland.co.uk/panels/488/) |
| R59 | Early onset or syndromic epilepsy | [R59 Genetic epilepsy syndromes](https://panelapp.genomicsengland.co.uk/panels/402/) |
| R61 | Childhood onset hereditary spastic paraplegia | [R61 Hereditary spastic paraplegia - childhood onset](https://panelapp.genomicsengland.co.uk/panels/568/) |
| R69 | Hypotonic infant with a likely central cause | [R69 Hypotonic infant](https://panelapp.genomicsengland.co.uk/panels/490/) |
| R83 | Arthrogryposis | [R83 Arthrogryposis](https://panelapp.genomicsengland.co.uk/panels/258/) |
| R84 | Cerebellar anomalies | [R84 Hereditary ataxia and cerebellar anomalies - childhood onset](https://panelapp.genomicsengland.co.uk/panels/488/) |
| R85 | Holoprosencephaly - NOT chromosomal | [R85 Holoprosencephaly](https://panelapp.genomicsengland.co.uk/panels/78/) |
| R86 | Hydrocephalus | [R86 Hydrocephalus](https://panelapp.genomicsengland.co.uk/panels/179/) |
| R87 | Cerebral malformation | [R87 Cerebral malformations](https://panelapp.genomicsengland.co.uk/panels/491/) |
| R88 | Severe microcephaly | [R88 Severe microcephaly](https://panelapp.genomicsengland.co.uk/panels/162/) |
| R98 | Likely inborn error of metabolism | [R98 Likely inborn error of metabolism](https://panelapp.genomicsengland.co.uk/panels/467/) |