## Appendix 3:

**Selected causes of microcephaly**

| Isolated microcephaly |
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| Autosomal recessive (multiple types) |
| Autosomal dominant |
| X-linked microcephaly |
| **Chromosomal abnormalities and syndromes** |
| Trisomies (eg, 21, 18, 13) |
| Monosomy 1p36 deletion |
| Seckel syndrome |
| Smith-Lemli-Opitz syndrome |
| Williams-Beuren syndrome (7q11.23 deletion) |
| Cornelia de Lange syndrome |
| Miller-Dieker lissencephaly syndrome (17p13.3 deletion) |
| Wolf-Hirschhorn syndrome (4p deletion) |
| Cri-du-chat syndrome (5p15.2 deletion) |
| Mowat-Wilson syndrome |
| Rubinstein-Taybi syndrome |
| Aicardi-Goutières syndrome |
| Cockayne syndrome |
| Bloom syndrome |
| Angelman syndrome |

| Neuroanatomic abnormalities associated with microcephaly |
|---|
| Neural tube defects (eg, anencephaly, hydranencephaly, encephalocele) |
| Holoprosencephaly |
| Atelencephaly (aprosencephaly) |
| Lissencephaly |
| Schizencephaly |
| Polymicrogyria |
| Pachygyria (macrogyria) |
| Fetal brain disruption sequence |
| **Metabolic disorders** |
| Maternal diabetes mellitus |
| Untreated maternal phenylketonuria |
| Phenylketonuria |
| Methylmalonic aciduria |
| Citrullinemia |
| Neuronal ceroid lipofuscinosis |
| **Environmental causes** |
| Congenital infection (eg, cytomegalovirus, herpes simplex virus, rubella, varicella, toxoplasmosis, HIV, syphilis, enterovirus, Zika virus) |
| Meningitis |
| In utero drug or toxin exposure (eg, alcohol, tobacco, marijuana, cocaine, opioid, antineoplastic agents, antiepileptic agents, radiation, toluene) |
| Perinatal insult (eg, hypoglycemia, hypothyroidism, hypopituitarism, hypoadrenalism) |
| Anoxia/ischemia |

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