

Appendix S2

Supplemental Table 2. Grade of severity for neurofibromatosis 1 by Riccardi

Stage	Clinical symptoms	
1	minimal	Café au lait spots only, or with unobtrusive cutaneous neurofibromas
2	mild	Numerous cutaneous neurofibromas but without facial disfigurement Small plexiform neurofibromas with no associated problems Asymptomatic osseous lesions Learning difficulties with normal IQ
3	moderate	Numerous cutaneous neurofibromas with facial disfigurement Plexiform neurofibromas with modest localised hypertrophy Visceral neurofibromas Mild intellectual disability Scoliosis or pseudoarthrosis requiring surgery Controlled epilepsy
4	severe	Disease complications leading to major health impairment, often requiring surgical intervention (Large plexiform neurofibromas with severe secondary problems, CNS tumors, malignancy, aqueduct stenosis, severe intellectual disability, pheochromocytoma, renal artery stenosis)