Supplementary Methods

cDNA constructs and cell culture experiments

CSF-1R cDNA (NM_005211) was amplified by PCR using a human whole-brain Marathon-Ready cDNA library (Clontech) as a template. Mutant CSF-1R cDNAs were generated using the Gene-Art site-directed mutagenesis system (Invitrogen). The deletion type of mutants (aberrant splice variants (ASVs) 1-3) associated with the splice-site mutation (c.2442+1G>T) were amplified by the specific primers using the cDNA obtained from brain of the patient. The primer sequences are available on request. To establish inducible cell lines that express wild-type (WT), the CSF-1R tagged with the FLAG was cotransfected into Flp-In T-Rex HEK293 cells. Hygromycin-resistant colonies were selected, and transgene expression was induced with 1 μg/mL doxycyclin (Sigma-Aldrich). Recombinant human M-CSF (R&D System) and IL-34 (R&D System) were used at a final concentration of 25 ng/mL.