# *RELA* fusion (Non-core)

**Reason/Evidentiary Support**

Approximately two-thirds of supratentorial ependymomas in children are characterised by fusions between *C11orf95* and the *RELA* genes.[1,2] Detection of these fusions is essential for making the diagnosis of ependymoma, *RELA* fusion positive. These fusions can be identified clinically using RNA sequencing, RT-PCR based techniques, or FISH; whole genome sequencing can also detect the fusion. Targeted RNA sequencing and RT-PCR design should take into consideration the complex nature of the fusion events generated by chromothripsis on chromosome 11. FISH probes overlying either *RELA* or *C11orf95* may be used to detect the rearrangements on chromosome 11.[1] These are designed using a break-apart strategy with red and green probes lying close to one another and producing a yellow signal in the wildtype situation; rearrangements will result in distancing of the probes from one another and distinct red and green signals. There are correlations between the presence of L1CAM positivity and RELA fusion in this type of this tumour (see **L1CAM expression (immunohistochemistry)**). There may also be other surrogate markers for *RELA* fusion–positive tumours and therefore other validated equivalents can be used to guide diagnosis; however, to date none of these is specific for *RELA* fusion as defined by FISH or sequencing.

**References**


1 Parker M, Mohankumar KM, Punchihewa C, Weinlich R, Dalton JD, Li Y, Lee R, Tatevossian RG, Phoenix TN, Thiruvenkatam R, White E, Tang B, Orisme W, Gupta K, Rusch M, Chen X, Li Y, Nagahawhatte P, Hedlund E, Finkelstein D, Wu G, Shurtleff S, Easton J, Boggs K, Yergeau D, Vadodaria B, Mulder HL, Becksfort J, Gupta P, Huether R, Ma J, Song G, Gajjar A, Merchant T, Boop F, Smith AA, Ding L, Lu C, Ochoa K, Zhao D, Fulton RS, Fulton LL, Mardis ER, Wilson RK, Downing JR, Green DR, Zhang J, Ellison DW and Gilbertson RJ (2014). C11orf95-RELA fusions drive oncogenic NF-kappaB signalling in ependymoma. *Nature* 506(7489):451-455.

2 Pajtler KW, Witt H, Sill M, Jones DT, Hovestadt V, Kratochwil F, Wani K, Tatevossian R, Punchihewa C, Johann P, Reimand J, Warnatz HJ, Ryzhova M, Mack S, Ramaswamy V, Capper D, Schweizer L, Sieber L, Wittmann A, Huang Z, van Sluis P, Volckmann R, Koster J, Versteeg R, Fults D, Toledano H, Avigad S, Hoffman LM, Donson AM, Foreman N, Hewer E, Zitterbart K, Gilbert M, Armstrong TS, Gupta N, Allen JC, Karajannis MA, Zagzag D, Hasselblatt M, Kulozik AE, Witt O, Collins VP, von Hoff K, Rutkowski S, Pietsch T, Bader G, Yaspo ML, von Deimling A, Lichter P, Taylor MD, Gilbertson R, Ellison DW, Aldape K, Korshunov A, Kool M and Pfister SM (2015). Molecular Classification of Ependymal Tumors across All CNS Compartments, Histopathological Grades, and Age Groups. *Cancer Cell* 27(5):728-743.