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REFERENCES

1. **Evans DG**, Mason S, Huson SM, Ponder M, Harding AE, Strachan T. Spinal and cutaneous schwannomatosis is a variant form of type 2 neurofibromatosis: a clinical and molecular study. *J Neural Neurosurg Psychiatry* 1997;**62**:361–6.
2. **Taylor MD**, Gokgoz N, Andrus IL, Mainprize TG, Drake JM, Rutka JT. Familial posterior fossa brain tumors of infancy secondary to germline mutation of the hSNF5 gene. *Am J Hum Genet* 2000;**66**:1403–6.
3. **MacCollin M**, Willett C, Heinrich B, Jacoby LB, Acierno JS Jr, Perry A, Louis DN. Familial schwannomatosis: exclusion of the NF2 locus as the germline event. *Neurology* 2003;**60**:1968–74.
4. **Janson K**, Nedzi LA, David O, Schorin M, Walsh JW, Bhattacharjee M, Pridjian G, Tan L, Judkins AR, Biegel JA. Predisposition to atypical teratoid/rhabdoid tumor due to an inherited INI1 mutation. *Pediatr Blood Cancer* 2006;**47**:279–84.
5. **Ammerlaan AC**, Ararou A, Houben MP, Baas F, Tijssen CC, Teepeen JL, Wesseling P, Hulsebos TJ. Long-term survival and transmission of INI1-mutation via nonpenetrant males in a family with rhabdoid tumour predisposition syndrome. *Br J Cancer* 2008;**98**:474–9.
6. **Weeks DA**, Beckwith JB, Mierau GW, Luckey DW. Rhabdoid tumor of kidney. A report of 111 cases from the National Wilms' Tumor Study Pathology Center. *Am J Surg Pathol* 1989;**13**:439–58.
7. **Rorke LB**, Packer RJ, Biegel JA. Central nervous system atypical teratoid/rhabdoid tumors of infancy and childhood: definition of an entity. *J Neurosurg* 1996;**85**:56–65.
8. **Sevenet N**, Sheridan E, Amram D, Schneider P, Handgretinger R, Delattre O. Constitutional mutations of the hSNF5/INI1 gene predispose to a variety of cancers. *Am J Hum Genet* 1999;**65**:1342–8.
9. **Roberts CW**, Orkin SH. The SWI/SNF complex – chromatin and cancer. *Nat Rev Cancer* 2004;**4**:133–42.
10. **Versteeg I**, Sevenet N, Lange J, Rousseau-Merck MF, Ambros P, Handgretinger R, Aurias A, Delattre O. Truncating mutations of hSNF5/INI1 in aggressive paediatric cancer. *Nature* 1998;**394**:203–6.
11. **Biegel JA**, Tan L, Zhang F, Wainwright L, Russo P, Rorke LB. Alterations of the hSNF5/INI1 gene in central nervous system atypical teratoid/rhabdoid tumors and renal and extracranial rhabdoid tumors. *Clin Cancer Res* 2002;**8**:3461–7.
12. **MacCollin M**, Chiocca EA, Evans DG, Friedman JM, Horvitz R, Jaramillo D, Lev M, Mautner VF, Niimura M, Plotkin SR, Sang CN, Stemmer-Rachamimov A, Roach ES. Diagnostic criteria for schwannomatosis. *Neurology* 2005;**64**:1838–45.
13. **Hulsebos TJ**, Plomp AS, Wolterman RA, Robanus-Maandag EC, Baas F, Wesseling P. Germline mutation of INI1/SMARCB1 in familial schwannomatosis. *Am J Hum Genet* 2007;**80**:805–10.
14. **Sestini R**, Bacci C, Provenzano A, Genuardi M, Papi L. Evidence of a four-hit mechanism involving SMARCB1 and NF2 in schwannomatosis-associated schwannomas. *Hum Mutat* 2008;**29**:227–31.
15. **Hadfield KD**, Newman WG, Bowers NL, Wallace A, Bolger C, Colley A, McCann E, Trump D, Prescott T, Evans DG. Molecular characterisation of SMARCB1 and NF2 in familial and sporadic schwannomatosis. *J Med Genet* 2008;**45**:332–9.
16. **Boyd C**, Smith M, Kluwe L, Balogh A, Maccollin M, Plotkin S. Alterations in the SMARCB1 (INI1) tumor suppressor gene in familial schwannomatosis. *Clin Genet* 2008;**74**:358–66.
17. **Jacoby LB**, Jones D, Davis K, Kronn D, Short MP, Gusella J, MacCollin M. Molecular analysis of the NF2 tumor-suppressor gene in schwannomatosis. *Am J Hum Genet* 1997;**61**:1293–302.
18. **Chen JM**, Chuzhanova N, Stenson PD, Ferec C, Cooper DN. Meta-analysis of gross insertions causing human genetic disease: novel mutational mechanisms and the role of replication slippage. *Hum Mutat* 2005;**25**:207–21.
19. **Biegel JA**, Zhou JY, Rorke LB, Stenstrom C, Wainwright LM, Fogelgren B. Germ-line and acquired mutations of INI1 in atypical teratoid and rhabdoid tumors. *Cancer Res* 1999;**59**:74–9.
20. **Patil S**, Perry A, Maccollin M, Dong S, Betensky RA, Yeh TH, Gutmann DH, Stemmer-Rachamimov AO. Immunohistochemical analysis supports a role for INI1/SMARCB1 in hereditary forms of schwannomas, but not in solitary, sporadic schwannomas. *Brain Pathol* 2008;**18**:157–9.
21. **Betz BL**, Strobeck MW, Reisman DN, Knudsen ES, Weissman BE. Re-expression of hSNF5/INI1/BAF47 in pediatric tumor cells leads to G1 arrest associated with induction of p16ink4a and activation of RB. *Oncogene* 2002;**21**:5193–203.
22. **Zhang ZK**, Davies KP, Allen J, Zhu L, Pestell RG, Zagzag D, Kalpana GV. Cell cycle arrest and repression of cyclin D1 transcription by INI1/hSNF5. *Mol Cell Biol* 2002;**22**:5975–88.
23. **Oruetebarria I**, Venturini F, Kekkarainen T, Houweling A, Zijderduijn LM, Mohd-Sarip A, Vries RG, Hoebe RC, Verrijzer CP. P16INK4a is required for hSNF5 chromatin remodeler-induced cellular senescence in malignant rhabdoid tumor cells. *J Biol Chem* 2004;**279**:3807–16.
24. **Tsikitis M**, Zhang Z, Edelman W, Zagzag D, Kalpana GV. Genetic ablation of Cyclin D1 abrogates genesis of rhabdoid tumors resulting from INI1 loss. *Proc Natl Acad Sci USA* 2005;**102**:12129–34.

Correction

The letter by Rossi *et al* that was published in the November 2008 issue of the journal (Rossi E, Messa J, Zuffardi O. Ring syndrome: still true? *J Med Genet* 2008;**45**:766–8) should have been published in the same issue as the letter to which it was responding (Zollino M, Murdolo M, Neri G. The terminal 760 kb region on 4p16 is unlikely to be the critical interval for growth delay in Wolf–Hirschhorn syndrome. *J Med Genet* 2008;**45**:544). The journal apologises for this error.

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