| ***NF1* mutation analysis: dermal Schwann cells** | | | | |
|---|---|---|---|---|
| **Class** | **Cell culture** | **Gender** | **Germline mutation** | **Somatic mutation** |
| Class 1 | ADN1N -/- | male | c.1062+1G>A | LOH |
| Class 1 | RMN9N -/- | female | unknown | LOH |
| Class 2 | AIBC2N -/- | female | unknown | unknown |
| Class 2 | SCC5N -/- | female | c.4309G>T | LOH |
| Class 2 | SCC7N -/- | female | c.4309G>T | LOH |
| Class 2 | ABB2N -/- | female | complete NF1 deletion | unknown |
| Class 2 | ERS1N -/- | female | unknown | unknown |
| Class 2 | JML3N -/- | female | unknown | LOH |
| Class 2 | ABC8N -/- | female | c.1721+3G>A | unknown |
| Class 2 | CLT6N +/- | male | c.1754delTAAC | LOH |
| Class 2 | MGF33N +/- | male | c.4537C>T | unknown |
| ***NF1* mutation analysis: plexiform Schwann cells** | | | | |
| **Class** | **Culture (pNF)** | **Gender** | **Germline mutation*** | **Somatic mutation** |
| Class 1 | 4.7 | female | unknown | unknown# |
| Class 1 | 5.4 | female | unknown | unknown# |
| Class 2 | 5.5 | male | c.3456_3457 insA | LOH |
| Class 1 | 3.3 | male | c.4269 G>A (skip ex24, in-frame) | unknown |
| Class 2 | 4.4 | female | c.6709 C>T (R2237X) | del ex20-28 |
| Class 2 | 5.3 | female | unknown | c.5222 C>T (R1748X) |
| Class 2 | 0.13 | female | c.2252-1G>C (skip ex14, fs) | unknown |
| Class 1 | 0.6 | female | unknown | unknown# |
| Class 2 | 95.6 | male | c.2245 C>T (R816X) | c.6709 C>T (R2237X) |
| Class 2 | 95.1 | female | unknown | c.380delG |
| Class 2 | 97.9 | female | c.7259+1 G>T (skip ex40, in frame) | unknown# |

**Supplementary Table 10.** NF1 Mutational Analysis. We restricted somatic mutation analysis in dNFs to LOH analysis (21), comparing DNA from blood and dNF from the same patient. We used microsatellite markers located within or surrounding the NF1 gene for genotyping with ABI 377 and 3130*x* Genetic Analyzers. We also analyzed DNA from

d*NF1*-/- Schwann cell cultures from tumors exhibiting LOH. Germline mutations of patients with dNFs were identified by cDNA-SSCP-heteroduplex as described (22). We screened genomic DNA from tissue for which ample high-quality DNA was available for *NF1* gene mutations by denaturing high-performance liquid chromatography based heteroduplex analysis in 13 / 23 solid tumor samples (dermal neurofibromas (n = 10), plexiform neurofibromas (n = 3) and MPNSTs (n = 1)), using the WAVE analysis system (Transgenomic; Omaha, NE). We designed primers to reduce homology to *NF1* pseudogenes sequences, and employed MLPA and sequencing of RT-PCR products (23). We also characterized mutations in 8 / 11 plexiform Schwann cell cultures, using these methods. There was no clear association between mutation and tumor type. The MPNST harbored a germline *NF1* point mutation and a large deletion including the *NF1* locus as the somatic mutation. A single MPNST (not shown) harbored a germline *NF1* point mutation and a large deletion including the *NF1* locus as the somatic mutation.