

S12 Table: Down-regulated genes from the MPNST vs. NF signature previously related with NF and/or MPNST.

Rank	ENSGENE	hgnc_symbol	chrom_name	band	Relationship with NF or MPNST	Reference
235	ENSG00000188580	NKAIN2	6	q22.31	Deletion of its chromosomal region in NF1-associated cutaneous neurofibromas.	[1]
218	ENSG00000196154	S100A4	1	q21.3	Immunostaining reaction variable in malignant melanoma, and benign and malignant peripheral nerve sheath tumors.	[2]
208	ENSG00000197956	S100A6	1	q21.3	Its expression differentiates desmoplastic melanomas from MPNSTs but its diagnostic use is limited by overlapping. Differential expression in malignant melanoma, and benign and malignant peripheral nerve sheath tumors.	[3] [2]
163	ENSG00000197971	MBP	18	q23	Overexpressed in NF1-associated compared to sporadic MPNST. One of the proteins complexed in RNA granules with NF1:LRPPRC.	[4] [5]
147	ENSG00000152061	RABGAP1L	1	q25.1	Deletion of its chromosomal region in NF1-associated cutaneous neurofibromas.	[1]
139	ENSG00000105855	ITGB8	7	p21.1	MPNST-specific copy number variations.	[6]
129	ENSG00000168329	CX3CR1	3	p22.2	Chemokine receptor expressed from high to moderate levels in benign and malignant nerve sheath tumors.	[7]
128	ENSG00000107562	CXCL12	10	q11.21	High to moderate levels of expression in benign and malignant nerve sheath tumors. Chemokine promoting cell proliferation in MPNSTs.	[7] [8]
123	ENSG00000277632	CCL3	17	q12	Probably involved in recruitment of inflammatory cells by tumorigenic cells that could play a role in the auto-degeneration of Schwann cell tumors.	[9]
72	ENSG00000109099	PMP22	17	p12	Down-regulated in MPNST cell lines compared to normal Schwann cells.	[10]
63	ENSG00000261857	MIA	19	q13.2	Potential serum biomarker for tumor load in NF1.	[11]
59	ENSG00000158887	MPZ	1	q23.3	Down-regulated Schwann cell-specific gene in the comparison MPNST vs. pNF.	[12]
56	ENSG00000169554	ZEB2	2	q22.3	Transcription factor associated to the epithelial-mesenchymal transition in neurofibromatosis type 1, up-regulated in NF1-associated neurofibromas and Schwann cells.	[13]
42	ENSG00000189058	APOD	3	q29	Its expression increases with the formation of NFs and is down-regulated during malignant transformation of NFs.	[14]
41	ENSG00000189058	TIMP4	3	p25.2	Down-regulated in MPNST vs. pNF	[12]

					comparison.	
33	ENSG00000198910	L1CAM	X	q28	Down-regulated in MPNST vs. pNF comparison. Overexpressed in NF1-associated vs. sporadic MPNSTs. Highly expressed in benign NF1 tumors and down-regulated in MPNST vs. benign NF1-associated tumors.	[12] [4] [15]
28	ENSG00000065361	ERBB3	12	q13.2	Its expression is rare in NF1-related tumors, particularly in MPNST. Down-regulated in MPNST vs. pNF comparison. Variable expression in NFs and MPNSTs. It is present in MPNST invadopodia.	[16] [12] [17] [18]
23	ENSG00000132470	ITGB4	17	q25.1	Down-regulated in MPNST vs. pNF comparison. Its genomic region was amplified; copy number changes of this gene were observed in 32% of MPNSTs. Included in a significant gain aberration.	[12] [19] [20]
21	ENSG00000101265	RASSF2	20	p13	Down-regulated in MPNST vs. pNF comparison. Included in a significant deletion aberration.	[12] [20]
18	ENSG00000100146	SOX10	22	q13.1	Down-regulated in MPNST vs. pNF comparison. Down-regulated in comparison MPNST vs. Schwann cells. More specific than S100 in diagnosis of neural crest-derived tumors. Its complete silencing discriminates MPNSTs and cellular schwannomas.	[12] [10] [21] [22]
9	ENSG00000168477	TNXB	6	p21.32	Down-regulated in MPNST vs. pNF comparison.	[12]
5	ENSG00000071991	CDH19	18	q22.1	Significantly up-regulated in NF1 tumors. Potential role in dermal neurofibroma initiation.	[23]
2	ENSG00000160307	S100B	21	q22.3	Down-regulated in MPNST vs. pNF comparison. Diffuse staining in malignant melanoma and benign peripheral nerve sheath tumors; it reacts in 30% of MPNSTs.	[12] [2]

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