

Genetic Mutations and Associated Conditions

#	Gene / Mutation	Associated Condition(s)	Notes
1	GNAS1	Fibrous Dysplasia	Affects cAMP-mediated osteoblast differentiation
2	HLA-B27	Ankylosing Spondylitis, Seronegative Spondyloarthropathies	Strong genetic association
3	CTNNB1 (β-catenin)	Deep Fibromatoses (Desmoid Tumor)	Activates Wnt signaling pathway
4	APC	Familial Adenomatous Polyposis (FAP), Gardner Syndrome, Deep Fibromatoses	Tumor suppressor gene; activates Wnt pathway
5	MYH9-USP6 (t(17;22))	Nodular Fasciitis	Fusion gene; indicates clonal neoplastic origin
6	T(1;2)(p13;q37)	Tenosynovial Giant Cell Tumor (PVNS)	Affects collagen type VI α3
7	MDM2 amplification (Chr 12)	Well-Differentiated Liposarcoma	Common in retroperitoneal tumors
8	T(12;16)	Myxoid Liposarcoma	Translocation characteristic of this subtype
9	Complex Karyotype	Pleomorphic Liposarcoma	No specific mutation; multiple chromosomal abnormalities
10	T(X;18)(p11;q11)	Synovial Sarcoma	Fusion gene SS18; detected by FISH
11	Fumarate Hydratase (Chr 1q42.3)	Leiomyoma	Low diagnostic value; seen in some cases
12	BRAF / RAS	Benign Nevus, Dysplastic Nevus, Melanoma	Early mutations in melanocytic tumors
13	TP53 / PTEN / TERT	Melanoma (advanced stages)	Late mutations during malignant progression
14	TP53	Actinic Keratosis, SCC, Basal Cell Carcinoma	Common UV-induced mutation
15	FGFR3	Seborrheic Keratosis	Benign skin tumor; different pathogenesis
16	PTCH1	Basal Cell Carcinoma, Gorlin Syndrome (Basal Cell Nevus Syndrome)	Hedgehog pathway mutation