

Mutations/syndromes	Diagnosis
GNAS1, Mazabraud syndrome, Milanese-Albert syndrome	Fibrous dysplasia (FD)
VI collagen a-3, T(1;2)(p13q;37)	Tenosynovial giant cell tumour
High level of RANKL	Giant cell tumour
NF1, Gardner syndrome, Li-Fraumeni syndrome, Osler-Webber-Rendu syndrome	Soft tissue tumours
T(x:18)(p11;q11.2)	Synovial sarcoma
T(11;22)	Ewing sarcoma
MDM2	WD liposarcoma
T(12;16)	Myxoid
T(17;22), MYH9-USP6	Nodular fasciitis
CTNB1 (b-catenin), APC gens	Deep fibromatosis
Fumarate hydrates	Leiomyoma
Tp53(DNA)	Actinic keratosis

FGFR3	Seborrheic keratosis
PTCH1,TP53,Grolin syndrome	Basal cell carcinoma
BRAF,RAS	Nevus , first mutation in malenoma
DNA telomerase	Dysplastic nevus
Loss of p16	Malenoma (vertical growth)
Loss of p53 & PTEN	Metastatic malenoma

Done by: Hussam Daraghmeh