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Chromosome banding analyses of cells from fine needle aspirate biopsies from soft tissue and bone tumors. Is it clinically meaningful?

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Abstract

Morphologic evaluation of samples from fine needle aspiration (FNA) and core needle (CN) biopsies is an important part of the pretreatment diagnosis of bone and soft tissue tumors. As most such tumors have characteristic, sometimes even specific, chromosomal rearrangements, ancillary genetic analyses could provide important diagnostic information. Whereas directed analyses, such as fluorescence in situ hybridization or RT-PCR, for specific genetic aberrations are well suited for the relatively small cell numbers obtained with FNA biopsies, the possibility to obtain tumor karyotypes after cell culturing has been less well studied. In the present study, karyotypes from 114 FNA biopsies were compared to those in corresponding surgical tumor (ST) biopsies; in addition, results on 31 CN biopsies, and their corresponding tumor biopsies, were available. Of the 138 ST biopsies, 88 (64%) showed clonal acquired chromosome aberrations, 42 (30%) displayed a normal karyotype, and 8 (6%) did not yield any karyotype due to infection or poor cell growth.The corresponding figures for the 114 FNA biopsies were 27 (24%), 28 (25%), and 59 (52%), and for the 31 CN biopsies 15 (48%), 10 (32%), and 6 (19%). The relatively low success rate, with the possible exception of primitive round cell/Ewing sarcomas (abnormal karyotype in 6/11 FNA biopsies), strongly indicates that it is not meaningful to attempt cell culturing and chromosome banding analysis on cells from FNA biopsies in the management of patients with suspected bone or soft tissue tumors. The use of ancillary techniques such as fluorescence in situ hybridization (FISH) might however improve the diagnostic value from FNA biopsies. Our preliminary data suggest that if a pre-treatment karyotype is wanted, the cytogenetic analysis should be made on cells from CN biopsies, close to half of which showed an aberrant karyotype.

Introduction

Fine needle aspiration (FNA) and core needle (CN) biopsies are useful instruments in diagnosing soft tissue and bone tumors [1]. Both needle techniques are in most cases easy to perform, cost effective and well tolerated by the patient [2]. In Scandinavia, the further management of the patient is decided upon the pre-treatment diagnosis made on tumor cells from FNA and/or tissue from CN biopsies, combined with clinical and radiographic information [3]. Apart from the morphologic evaluation of tumor cells in FNA and CN biopsies, ancillary techniques, such as immunocytochemistry/immunohistochemistry, are often used to improve the diagnostic precision. As several bone and soft tissue tumors are characterized by specific genetic changes, notably translocations resulting in fusion genes, also the genetic status of the tumor can provide clinically useful information [4]. To ensure robust and rapid results, these genetic abnormalities are usually investigated by fluorescence in situ hybridization (FISH) or reverse transcriptase polymerase chain reaction (RT-PCR) analysis. A drawback with both techniques is that they provide information only on the abnormality that is assessed, i.e., they can support or exclude a tentative diagnosis, but rarely suggest an alternate diagnosis. Furthermore, most bone and soft tissue tumors do not have any known specific genetic changes, but display characteristic combinations of numerical and unbalanced structural chromosome aberrations. For instance, if the cytologic examination cannot decisively settle whether a lesion is benign or malignant, the finding of a near-triploid chromosomes number with multiple structural and numerical changes strongly argues for the latter. Thus, the genomic screening obtained through chromosome banding analysis of cultured cells could provide further diagnostic information.

Most of the cytogenetic information on bone and soft tissue tumors, presently amounting to data on close to 3,000 cases [5], comes from studies on ST biopsies, and very few attempts have been made to evaluate in larger series the possibility to obtain tumor karyotypes from needle biopsies [6- 8]. In the present study, we have assessed the results from G-banding analysis of 114 FNA biopsies, and compare the results with those from the corresponding ST biopsies. In addition, we also compared with preliminary data on 31 CN biopsies.

Materials and Methods

The study was based on fresh FNA (n=114), CN (n=31) and ST (n=138) biopsies obtained from a total of 139 patients with a bone or soft tissue tumor treated at the tumor orthopedic centre, Lund University Hospital, Lund, Sweden, between 1987 and 2009. Clinical information, histopathologic diagnoses and tumor karyotypes are given in Tables 1 and 2.

The FNA procedures were performed with 22–24 gauge needles attached to disposable, 10-mL syringes in a Cameco syringe holder (Cameco AB Sandviken, Sweden). The aspirates were air dried, stained with May–Grünwald–Giemsa (MGG), fixed in 95% ethanol, and stained with hematoxylin and eosin (H&E). The number of passes varied from 1 to 6 with an average of 3. Between 1987 and 1997 no separate passes for cytogenetic analysis were performed and the material derived from needle rinsing. From 1997, one separate pass for cytogenetic material was performed. Other changes introduced during the sampling period included trials with cellblocks between 1995 and 1997; since 1997, the cellblock technique is routine. In 2002, liquid-based immunocytology was introduced as a routine diagnostic method. The

sampling was performed by the cytopathologist in the majority of the cases. However, sampling of non-palpable lesions was performed in collaboration between the cytopathologist and a radiologist using image guidance, usually ultrasound. Rapid evaluation of the adequacy of the material with quick staining (DiffQuick or H&E) started in 1997 with selected cases and became routine in 2000. Concerning the material sent for cytogenetic analysis no specific rapid evaluation of the quantity or quality was performed. The CNB procedures were performed using an 18-gauge needle and the Pro-Mag™ 1,4 and 2,5 Automatic Biopsy Instrument. After washing the skin with 70% ethanol, local anesthesia was achieved by subcutaneous infiltration of 1–3 mL 1% lidocaine.

Short-term cell culturing, harvesting, and G-banding of chromosomes were performed as described [9], and the karyotypes were interpreted and written according to ISCN (2009)[10]. The cytogenetic analysis was considered successful if a clonal acquired aberration was detected.

Results

Of the 138 ST biopsies, 88 (64%) showed clonal acquired chromosome aberrations, 42 (30%) displayed a normal karyotype, and 8 (6%) did not yield any karyotype due to infection or poor cell growth. The corresponding figures for the 114 FNA biopsies were 27 (24%), 28 (25%), and 59 (52%), and for the 31 CN biopsies 15 (48%), 10 (32%), and 6 (19%) (Tables 3 and 4). When comparing benign and malignant tumors (here defined as tumors labeled sarcomas), abnormal karyotypes were seen in 14% and 28% of FNA samples, respectively (Table 5). Of the 27 FNA biopsies with clonal aberrations, 17 showed an identical or very similar karyotype in the ST biopsies, 3

displayed seemingly unrelated clones in the ST biopsies, 5 did not show any clonal changes in the ST biopsies, and in 2 cases there was no corresponding ST biopsies. Of the 15 CN biopsies with clonal aberrations, 8 showed an identical or very similar karyotype in the ST biopsies, 2 displayed seemingly unrelated clones in the ST biopsies, 3 did not show any clonal changes in the ST biopsies, and for 2 samples there was no corresponding ST biopsies.

Discussion

The main aim of the present study was to evaluate the success rate for chromosome banding analysis of cells from FNA biopsies from bone and soft tissue tumors. For that purpose, karyotypes from 114 FNA biopsies from 111 patients were retrieved and compared with the results of ST biopsies from the corresponding tumors. The overall success rate for cytogenetic analysis on material provided from FNA was low in our material. Only in 24% of the samples was an abnormal karyotype identified, compared to 64% of the ST biopsy material. Nor were any major differences seen when subdividing the material according to the origin of the tumor, bone vs soft tissue, or whether they were benign or malignant (Tables 3-5). Thus, the results on FNA biopsies were consistently poor, CN biopsies showed better and ST biopsies the best results.

Bone and soft tissue tumors are histologically heterogeneous, and only for a few subgroups did the number of investigated cases allow for an attempt to compare the cytogenetic success rate with lineage of differentiation (Table 6). The only group of tumors that showed a notably higher fraction of abnormal karyotypes was Ewing sarcoma/primitive neuroectodermal tumors; an abnormal karyotype was found in 6,

including one case with a single abnormal cell, of the 11 FNA samples. This finding is in agreement with previous data [8]. The group that showed the lowest success rate and the highest failure rate was the adipose tissue tumors. This could be attributed to the characteristics of adipose tumor tissue which makes it difficult to aspirate adequate sample material. In spite of the relatively high success rate in Ewing tumors, one must conclude that chromosome banding analysis of cells from FNA biopsies is not a cost-effective method to search for genetic information of diagnostic relevance in the management of patients with bone and soft tissue tumors. This conclusion does not, of course, apply to the directed analyses. Both FISH and RT-PCR have been shown to be very useful when searching for specific neoplasia-associated rearrangements, such as fusion genes, in samples with small numbers of tumor cells [11, 12].

The reason(s) for the relatively poor success rate for chromosome banding analysis of FNA biopsies is not known, but the sheer quantity of tumor cells might be one factor. In support of this notion, the, admittedly preliminary, data on 31 CN biopsies suggest that already this relatively small increase in sample size results in a much higher success rate. An abnormal karyotype was found in 15 of 31 (48%) CN biopsies, to be compared with 24% of FNA biopsies and 64% of ST biopsies.

What other options, then, exist for obtaining a preoperative genomic tumor screening? Although array-based technologies for either global gene expression patterns or genomic imbalances are potentially very powerful screening methods, they are still used mainly as research tools, and their limitations with regard to the size and quality of the tumor samples have been poorly explored. Nevertheless, there are some promising results on FNA material from papillary thyroid carcinoma and non-small cell lung carcinomas [13-15]. The same techniques should be applicable

also to soft tissue and bone tumors, but further studies are needed to evaluate this. Still, the present study suggests that CN biopsies are more promising than FNA biopsies in this context. Also CN is well tolerated by the patient, cost effective and relatively easy to perform [2, 16]. In addition, CN biopsies provide better information on the architecture of the tumor tissue, as well as more reliable material for ancillary techniques such as immunohistochemistry, FISH and RT-PCR.

FNA can provide diagnostic smears for musculoskeletal lesions but as a standalone method for further evaluation it has limitations [17]. In our opinion, FNA and CN complement each other as diagnostic instruments for cytomorphologic evaluation, but when it comes to genetic screening through chromosome banding analysis CN seems superior to FNA. However, one should not forget the possibility to use ancillary techniques directly on FNA material, which can prove to be useful [18]. Furthermore, FNA and CN can be performed simultaneously by the same examiner, thus minimizing the time from examination to diagnosis and providing material for further diagnostic work up.

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References

[1] Verheijen P, Witjes H, van Gorp J, Hennipman A, van Dalen T. Current pathology work-up of extremity soft tissue sarcomas, evaluation of the validity of different techniques. Eur J Surg Oncol 2010;36:95-9.

[2] Layfield LJ. Cytologic diagnosis of osseous lesions: a review with emphasis on the diagnosis of primary neoplasms of bone. Diagn Cytopathol 2009;37:299-310.

[3] Dalén MBP, Meis-Kindblom JM, Sumathi VP, Ryd W, Kindblom LG. Fine needle aspiration and core needle biopsy in the preoperative diagnosis of desmoid tumors. Acta Orthop Scand 2006;77:926-31.

[4] Mertens F, Panagopoulos I, Mandahl N. Genomic characteristics of soft tissue sarcomas. Virchows Arch 2010;456:129-39.

[5] Mitelman Database of Chromosome Aberrations in Cancer 2010. Mitelman F, Johansson B and Mertens F (Eds.), http://cgap.nci.nih.gov/Chromosomes/Mitelman

[6] Molenaar WM, Van den Berg E, Dolfin AC, Zorgdrager H, Hoekstra HJ. Cytogenetics of fine needle aspiration biopsies of sarcomas. Cancer Genet Cytogenet 1995;84:27-31.

[7] Akerman M, Dreinhöfer K, Rydholm A, Willén H, Mertens F, Mitelman F, et al. Cytogenetic studies on fine-needle aspiration samples from osteosarcoma and Ewing´s sarcoma. Diagn Cytopathol 1996;15:17-22.

[8] Udayakumar AM, Sundareshan TS, Mallan Goud T, Gayathri Devi M, Biswas S, Appaji L, et al. Cytogenetic characterization of Ewing tumors using fine needle aspiration samples: a 10-year experience and review of the litterature. Cancer Genet Cytogenet 2001;127:42-8.

[9] Mandahl N. Methods in solid tumor cytogenetics, In: Rooney DE, editor. Human Cytogenetics: Malignancy and Acquired Abnormalities, 3rd Ed. New York: Oxford University Press. 2001; pp.165-203.

[10] Shaffer LG, Slovak ML, Campbell LJ, editors. ISCN: an International System for Human Cytogenetic Nomenclature, Basel: S. Karger, 2009.

[11] Lazar A, Abruzzo LV, Pollock RE, Lee S, Czerniak B. Molecular diagnosis of sarcomas: chromosomal translocations in sarcomas. Arch Pathol Lab Med 2006;130:1199-1207.

[12] Tanas MR, Goldblum JR. Fluorescence in situ hybridization in the diagnosis of soft tissue neoplasms: a review. Adv Anat Pathol 2009:16:383-391.

[13] Chen YT, Kitabayashi N, Zhou XK, Fahey III TJ, Scognamiglio T. MicroRNA analysis as a potential diagnostic tool for papillary thyroid carcinoma. Mod Pathol 2008; 21:1139-46.

[14] Lim EH, Aggarwal A, Agasthian T, Wong PS, Tan C, Sim E, et al. Feasibility of using low volume tissue samples for gene expression profiling of advanced non-small cell lung cancers. Clin Cancer Res 2003; 9:5980-87.

[15] Soglio DBD, Rougemont AL, Absi R, Barrette S, Montpetit A, Fetni R, et al. SNP genotyping of a sclerosing rhabdomyosarcoma: reveals highly aneuploid profile and a specific MDM2/HMGA2 amplification. Hum Pathol 2009; 40:1347-52.

[16] Chowdhury T, Barnacle A, Haque S, Sebire N, Gibson S, Andersson J, et al. Ultrasound-guided core needle biopsy for the diagnosis of rhabdomyosarcoma in childhood. Pediatr Blood Cancer 2009; 53:356-60.

[17] Domanski H, Akerman M, Carlen B, Engellau J, Gustafson P, Jonsson K, et al. Core-needle biopsy performed by the cytopathologist. A technique to complement fine-needle aspiration of soft tissue and bone lesions. Cancer 2005; 105:229-39.

[18] Sapi Z, Antal I, Papai Z, Szendroi M, Mayer A, Jakab K, et al. Diagnosis of soft tissue tumours by fine-needle aspiration with combined cytopathology and ancillary techniques. Diagn Cytopathol 2002; 26:232-42.

Table 1. Cytogenetic findings in cell cultures from fine needle aspiration biopsies and corresponding surgical tumor biopsies. Clinical data and karyotypes in 111 patients with soft tissue or bone tumors.

Age/sex	Type of sample[a]	Karyotype[b]	Diagnosis	Reference[c]
		Soft tissue tumors		
<u>Adipocytic tumors</u>				
35/M	FNA	Failure	Lipoma	-
	ST	46,XY,t(1;12)(p32;q14)[16]/46,XY[3]		
56/M	FNA	46,XY[25]	Lipoma	-
	ST	46,XY,der(11)del(11)(q13)t(11;?16)(p15;q22),der(16)t(11;16)(q13;q22)[18]/46,XY[7]		
45/M	FNA	**46,XY,?inv(5)(q14q22),t(5;12)(q31;q14)**[8]	Lipoma	
	ST	46,XY,?inv(5)(q14q22),t(5;12)(q31;q14)[22]		11858:89
57/M	FNA	**46,XY,t(2;12)(q36;q14)[**22]/**46,idem,t(1;7)(p32;p15)**[6]/**46,idem,t(1;2)(q31;q11)**[3]	Lipoma	
	ST	46,XY,t(2;12)(q36;q14)[54]		11858:31
19/M	FNA	Failure	Chondroid lipoma	

	ST	46,XY,t(1;2;5)(q32;q37;q31),t(11;16)(q13;p13)[24]		8418:1
60/M	FNA	Failure	Pleomorphic lipoma	-
	ST	46,XY[10]		
53/M	FNA	Failure	Spindle cell lipoma	
	ST	46,XY,t(4;6)(q25;p23),der(11)t(11;13)(p15;q14),del(13)(q12q13)[5]/		9986:35
		46,idem,tas(Y;21)(p11;p13)[3]		
78/M	FNA*	Failure	Spindle cell lipoma	-
	ST	45,XY,der(2)t(2;?13)(q13-21;q21-31),add(13)(q13),-16[16]		
76/M	FNA	**46,XY,del(13)(q14q22)**[25]	Spindle cell lipoma	
	ST	46,XY,inv(9)(p11q12)c,del(13)(q12q21)[11]		12980:48
1/F	FNA	46,XX[14]	Lipoblastoma	-
	ST	46,XX[22]		
0/F	FNA	Failure	Lipoblastoma	
	ST	46,XX,t(1;8)(p13;q11)[19]/46,XX[6]		12454:6
66/M	FNA	46,XY[16]	Atypical lipomatous tumor	
	ST	47,XY,+r[10]/46,XY[13]		10805:10
70/M	FNA	Failure	Atypical lipomatous tumor	

	ST	47,XY,+r[10]/48,idem,+r[3]		11858:209
49/M	FNA	46,XY[23]	Atypical lipomatous tumor	-
	ST	46-47,XY,+1-2r[cp17]/46,XY[5]		
76/M	FNA	Failure	Atypical lipomatous tumor	-
	ST	47-49,XY,add(11)(p15),+1-3r[cp9]		
78/F	FNA	Failure	Well differentiated liposarcoma	12975:10
	ST	44-46,XX,+2r[cp6]/90-94,idemx2[cp4]		
32/M	FNA	Failure	Well differentiated liposarcoma	-
	ST	46,XY		
55/F	FNA	Failure	Dedifferentiated liposarcoma	10805:14
	ST	47-49,XX,?add(7)(p22),add(12)(q13),-14,-16,+der(?)r(?;1;12)x1-3, +der(?)t(?;1;12) [cp7]/46,XX[5]		
59/F	FNA	Failure	Myxoid liposarcoma	-
	ST	46,XX		
55/F	FNA	Failure	Myxoid liposarcoma	-
	ST	46,XX,t(12;16)(q13;p11)[25]		
80/F	FNA	Failure	Sclerosing liposarcoma	

	ST	41-44,XX,der(2)t(2;12)(q37;q13),del(4)(p14),del(11)(q13q21),-12,-12,-13,-14,-17, -18,add(19)(q13),+21,+?add(21)(q22),der(22)t(12;22)(q13;q12),+der(?)t(?;1;12) [cp4]/82-88,idemx2[cp2]/46,XX[6]		10805:16
60/M	FNA*	**46,XY,t(12;13;16)(q13;q12;p11)**[21]/46,XY[6]	Myxoid liposarcoma	
	ST	46,XY,t(12;13;16)(q13;q12;p11**)**[25]		12980:72
76/M	FNA	Failure	Pleomorphic liposarcoma	
	ST	40-45,X,-Y,-3,-9,add(11)(p15),der(11)t(11;12)(p13;q12),der(11)t(3;11)(p21;p15), -12,der(14)t(9;14)(q13;p11),del(17)(q11q21),der(19)add(19)(p13)add(19)(q13), -21,+1-2r,+1-2mar[cp2]/77-95,XX,-Y,-Y,-1,-3,-3,-4,-5,-6,-7,-8,-8,-9,-9,-9,-10, add(11)x2,der(11)t(10;11)(q22;p15)x2,der(11)t(3;11)x2,-12,add(12)(p13),-13,-14, -15,-16,-17,del(17)x2,der(19)x2,-20,-21,-22,+2-5r,+2-14mar[cp8]/47-49,XY,add(11), +1-3r[cp9]/46-49,XY,add(11),der(11)t(11;13)(p12;q13),-13,+2-4r[cp9]		5609:44
Fibroblastic/myofibroblastic tumors				
70/F	FNA	Failure	Ischemic fasciitis and lipoma	-
	ST	47,XX,+r[6]/46,XX,t(5;8)(q13;p21)[9]		
30/M	FNA	Failure	Myositis ossificans	-
	ST	46,XY[24]		

36/M	FNA	**47,XY,+5,-21,der(22)t(?17;22)(q22;q13),+r**[25]	Dermatofibrosarcoma protuberans	-
	ST	47-50,XY,+5,-21,der(22)t(?17;22)(q22;q13),+1-4r[cp10]		
16/M	FNA	Failure	Desmoid type fibromatosis	
	ST	47,XY,+8[6]/46,XY[21]		9163:401
43/F	FNA	46,XX[25]	Desmoid type fibromatosis	-
	ST	46,XX,del(6)(q2?3),tas(20;21)(q13;p13),+mar[cp8]		
17/M	FNA	Failure	Solitary fibrous tumour	-
	ST	46,XY[23]		
53/F	FNA	46,XX[25]	Solitary fibrous tumour	-
	ST	46,XX[25]		
12/F	FNA	Failure	Inflammatory myofibroblastic tumor	
	ST	46,XX,der(2)t(2;8)(p21;q21)del(2)(q31q37),der(8)t(2;8)ins(8;2)(q21;q31q37)[17]		5609:54

45/M	FNA	**41-42,XY,dic(1;?)(p11;?),-2,der(3)t(3;10)(p21;q?),-9,add(10)(q2?4),del(10)(q?22), der(12)add(12)(p1?1)t(?;10)(?;q?11)t(1;10)(p?;q?22),add(14)(q32),?add(16)(p11), -17,add(21)(p11),-22,-22**[3]/46,XY[4]	Myxoinflammatory fibroblastic sarcoma	
	ST	41-42,XY,dic(1;?)(p11;?),-2,der(3)t(3;10)(p21;q?),-9,add(10)(q2?4),del(10)(q?22), der(12)add(12)(p1?1)t(?;10)(?;q?11)t(1;10)(p?;q?22),add(14)(q32),?add(16)(p11), -17,add(21)(p11),-22,-22[cp19]		12793:2
0/M	FNA	Failure	Infantile fibrosarcoma	
	ST	49,XY,+8,+11,+20[6]/50,idem,+17[9]/46,XY[13]		2983:1
78/M	FNA	Failure	Myxofibrosarcoma	-
	ST	37-43,XY,-1,-1,-2,?del(2)(p14),add(9)(q34),?der(10)t(1;10)(q2?1;q2?6),-13,-16, +?21,+?r,inc[cp10]		
82/M	ST	39-46,XY,add(1)(p11),-2,-9,?add(9)(q32),-10,add(13)(q22),add(14)(q1?3), add(15)(p11),der(17)t(1;17)(p1?1;p13)ins(17;?)(p13;?),-18,-20,inc[cp6]	Myxofibrosarcoma	-
	FNA[d]	**68-78,der(X)t(X;1)(p22;p22)x2,-Y,+2,add(2)(p23)x2,+5,+9,?der(9)add(9)(p13) t(9;17)(q22;q21)x2,-10,add(11)(q23),+add(11),del(11)(p13)x2,+14,?der(14;15) (q10;q10)x2,-17,-18,der(22)t(10;22)(q11;p13)x2,+mar,inc**[cp6]		
76/M	FNA	**43-47,XY,der(1)t(1;3)(p13;q11),-3,-4,-8,?del(9)(q11),der(10)t(1;?;10)(p13;?;q22),**	Myxofibrosarcoma	-

		der(12)t(1;12)(q11;p11),?der(12)t(12;17)(q13;q21),-13,-15,-16,+1-4r,inc[cp8]		
	ST	42-45,XY,der(1)t(1;3)(p13;q11),-3,add(5)(p1?5),add(8)(q24),?del(9)(p11),der(10) t(1;?;10)(p13;?;q22),der(12)t(1;12)(q11;p11),?der(12)t(12;17)(q13;q21),-13,-14,-15, -16,+1-3r,inc[cp8]/46,XY[12]		
64/M	FNA	Failure	Myxofibrosarcoma	-
	ST	78-154,X?,add(1)(q21),?der(4)add(4)(q?)hsr(?),add(11)(p15),del(12)(p11), add(13)(q34),inc[cp2]/46,XY[7]		
77/F	FNA	46,XX[25]	Myxofibrosarcoma	-
	FNA	Failure		
	ST	64-79,XXX,+1,add(1)(p36)x2,-2,del(3)(q27),-4,+7,?add(7)(q21),-9,?add(10)(q22)x2, ?del(10)(q24),-11,-12,-13,-13,-13,-14,-14,-14,-15,-15,-16,-16,-16,-17,?add(18)(p11), ?add(19)(q13)x2,+add(19)(p13),+20,-22,inc[cp4]		
72/F	FNA	Failure	Myxofibrosarcoma	-
	ST	Failure		
55/M	FNA	Failure	Myxofibrosarcoma	-
	ST	72-75,XY,-X,-1,dic(1;13)(p13;p13),-2,-2,-2,-3,?add(3)(p25)x 1-2,?del(4)(p11), del(5)(p11)x1-2,-6,-6,-7,del(7)(q27)x1-2,-8,-8,-9,-9,?del(9)(q13),-10,-10,-10,-11,		

		?add(12)(p11),-13,-15,add(15)(p11),-16,add(16)(q?)x1-2,?del(17)(p11),?-20, ?add(21)(q22),inc[cp5]/46,XY[2]		
77/F	FNA	46,XX[8]	Myxofibrosarcoma	-
	ST	46,XX[22]		
76/M	FNA	**40-44,XY,der(1;16)(q10;p10),-5,-6,-12,-13,ins(13;?)(q14;?),+add(16)(q22),-17,-19, del(20)(q11),-21,-22,+der(?)t(?;12)(?;q13-14),+2mar,inc**[cp6]/46,XY[6]	Myxofibrosarcoma	-
	ST[d]	42-43,XY,der(1;16)(q10;p10),add(6)(q15),ins(13;?)(q14;?),del(20)(q11),inc[4]		
44/M	FNA	Failure	Myxofibrosarcoma	-
	ST	44-45,XY,-6,-8,-9,-10,?inv(10)(p12q24),?dup(17)(q11q21),+2mar[cp7]		
71/M	FNA	**81-94,XX,-Y,-Y,-1,-1,-3,-3,-4,-4,-4,+7,add(7)(q31)x2,dup(11)(q12q25)x2,-13,-13,+19, +19,+add(19)(q13)x2,+20,+20,+20,+22,+3mar**[cp4]	Myxofibrosarcoma	-
	ST	99-113,XXX,-Y,-Y,del(1)(q32)x2,add(7)(q31)x2-3,dup(11)(q12q25)x2,add(19)(q13)x2, inc[cp4]		
39/M	FNA	46,XY[12]	Low grade fibromyxoid sarcoma	
	ST	46,XY,t(7;16)(q32;p11)[5]/46,XY[11]		10986:4
Smooth muscle tumors				
55/M	FNA	46,XY[25]	Leiomyosarcoma	

	FNA	46,XY[8]		
	ST	45,X,-Y[17]		8300:31
63/M	FNA	**76-91,XX,-Y,del(1)(q42),add(12)(q24),add(19)(p13),inc[cp5]**	Leiomyosarcoma	
	ST	45-51,XY,der(1)del(1)(p36)add(1)(q32),add(3)(p25),add(7)(p11),del(7)(q11q22), add(12)(q24),add(17)(q25),add(19)(q13),der(19)t(5;19)(q13;p11)[5]/45-49,idem, -add(3),+del(3)(q11)[6]/81-97,idemx2,-add(3)x2,+del(3)x2[4]/42-44,idem, add(1)(q32),del(4)(p14),add(5)(p15),-del(7),-add(19),-der(19)[4]		5609:40
79/F	FNA	46,XX[6]	Leiomyosarcoma	
	ST	64-67,XXX,add(2)(p15),del(2)(q23),add(7)(p13)x2,add(8)(p11),add(8)(p21),add(10) (q26),add(15)(q22),der(15)t(9;15)(q13;p13),der(18)t(11;18)(q14;p11),+der(?)t(?;3) (?;p13),+hsr(?),inc[cp7]/95-119,idemx2,del(1)(p32),der(1;14)(p10;q10),der(11;22) (q10;q10)[cp9]		7478:220
86/F	FNA	Failure	Leiomyosarcoma	
	ST	44,X,add(X)(p22),-10,-13,add(15)(q15),add(16)(q24),der(16)t(15;16)(q15;p13), add(17)(p11),-22,+r[7]/43,idem,-2[3]/45,idem,+8[6]/46,XX[2]		8300:3
70/M	FNA	Failure	Leiomyosarcoma	8300:7
	ST[d]	52-104,XY,add(1)(p36),add(1)(q12),del(1)(p11),add(2)(p11),add(3)(p21),add(5)		

		(p15),add(6)(p24),add(7)(p22),add(7)(q22),add(8)(p11),add(11)(p15),add(11)(q24), add(22)(q13),+hsr(?),inc[cp4]/46,XY[14]		
72/F	FNA	46,XX[21]	Leiomyosarcoma	-
	ST	46,XX,inv(7)(p21q11),-12,+der(?)t(?;12)(?;q15)[9]/45,X,-X[3]/46,XX[8]		
81/M	FNA	Failure	Leiomyosarcoma	-
	ST	63-77,X?,inc[4]/127-152,X?,inc[5]/46,XY[9]		
51/F	FNA	Failure	Leiomyosarcoma	-
	ST	46,XX[23]		
78/F	FNA	Failure	Leiomyosarcoma	-
	ST	Failure		
82/M	FNA	Failure	Leiomyosarcoma	-
	ST	45,X,-Y[5]/47,XY,+7[2]/46,XY[17]		
81/M	FNA	45,XY,der(13;14)(q10;q10)c[19]	Leiomyosarcoma	-
	ST	45,XY,der(13;14)(q10;q10)c[23]		
49/M	FNA	Failure	Leiomyosarcoma	
	ST	79-92,XXYY,add(1)(q21)x2,t(11;14)(q23;q32),-17,inc[cp7]		10805:37
73/M	FNA	**42-43,Y,add(X)(q28),-1,del(1)(q21),add(2)(p14),?del(3)(p11), der(3)t(3;?13)**	Leiomyosarcoma	-

		(p13;q12),der(5)t(5;15)(p15;q15),t(7;8)(p15;q13),-8,-9,-13,-15,-15,add(16)(q22),		
		der(17)t(1;17)(q12;q21),add(21)(p11),add(22)(q13),der(22)t(8;22)(q11;p11),		
		+2mar[cp7]		
	ST	41-43,Y,add(X)(q28),-1,del(1)(q21),add(2)(p14),?del(3)(p11),der(3)t(3;?13)		
		(p13;q12),der(5)t(5;15)(p15;q15),t(7;8) (p15;q13),-8,-9,-13,-15,-15,add(16)(q22),		
		der(17)t(1;17)(q12;q21),add(21)(p11),add(22)(q13),der(22)t(8;22)(q11;p11),		
		+2mar[24]/46,XY[11]		
45/F	FNA	Failure	Leiomyosarcoma	-
	ST	57-95,XX,-X,9-10dmin,inc[5]/46,XX[8]		
82/M	FNA	**46,X,-Y,+7[12]**	Leiomyosarcoma	-
	ST	39-49,XY,-3,+2-6r[cp7]/85-92,XXYY,+5-9r,inc[cp2]		
Pericytic (perivascular) tumors				
64/F	FNA	**72,X,-X,add(7)(p22),+der(?)t(?;1)(?;p11),inc**[2]/46,XX[13]	Hemangiopericytoma	8108:318
	ST	46,XX[25]		
84/M	FNA	Failure	Hemangiopericytoma	
	ST	46,XY,t(2;17)(p16;q11),ins(7;11)(q36;q14q23)[11]/46,XY[3]		8108:319
50/M	FNA	Failure	Hemangiopericytoma	-
	ST	46,XY[24]		

Skeletal muscle tumors

10/F	FNA	**118,XXXXX,+X,+X,add(1)(p11)x2,+2,-3,-4,-4,+5,+5,-6,-7,+8,+9,-10,-11,+12,-14,-16, +20,+20,+20,-21,+22,+der(?)t(?;5)(?;q15)x2[2]**	Alveolar rhabdomyosarcoma	7200:2
	ST	118,XXXXX,+X,+X,add(1)(p11)x2,+2,-3,-4,-4,+5,+5,-6,-7,+8,+9,-10,-11,+12,-14,-16, +20,+20,+20,-21,+22,+der(?)t(?;5)(?;q15)X2[2]/46,XX[16]		7200:2
15/M	FNA	Failure	Alveolar rhabdomyosarcoma	-
	ST	46,XY[21]		
3/F	FNA	46,XX[13]	Alveolar rhabdomyosarcoma	-
	ST	46,XX[24]		
9/F	FNA	Failure	Alveolar rhabdomyosarcoma	-
	ST	46,XX[17]		

Soft tissue tumors of miscellaneous/uncertain differentiation

57/F	FNA	**46,XX,t(1;10)(p21;q25)**[13]/46,XX[15]	Myxoma	-
	ST	46,XX[24]		
45/F	FNA	46,XX[12]	Myxoma	-
	ST	46,XX[25]		
81/F	FNA	Failure	Neurilemmoma	

	ST	47,XX,+20[6]/45,X,-X[3]/46,XX[28]		8439:6
12/F	FNA	46,XX[5]	Neurofibroma	-
	ST	46,XX[28]		
73/F	FNA	Failure	Malignant peripheral nerve sheath tumor	-
	ST	Failure		
68/M	FNA	**82-140<4n>,XY,-Y,-Y,+add(1)(q21)x2-3,add(6)(q11)x2,add(6)(q13)x2,add(8)(p11),-10,-10,add(11)(q13),del(12)(q24),add(14)(p11),-16,add(16)(p11),-17,-17,-18,-18,add(19)(p13)x2,+add(19)(p11),-20,-20,-20,add(22)(p13)x1-2,+der(?)t(?;7)(?;q11),inc**[cp6]	Undifferentiated pleomorphic sarcoma	-
	ST	73-88,XX,-Y,-Y,+add(1)(q21)x2,add(6)(q13)x2,?add(16)(q1?1),add(19)(p13),add(22)(p13)x2,+der(?)t(?;7)(?;q11),inc[cp3]/45,X,-Y[3]/46,XY[4]		
75/F	FNA	Failure	Undifferentiated pleomorphic sarcoma	
	ST	79-97,XX,-X,-X,add(1)(q11),del(1)(q12),add(7)(p22),der(16)add(16)(p13)hsr(?)x2,dmin,inc[cp4]/46,XX[?]		5609:38
79/F	FNA	46,XX[9]	Undifferentiated pleomorphic sarcoma	-
	ST	46,XX[30]		
78/F	FNA	Failure	Undifferentiated pleomorphic sarcoma	-

	ST	46,XX[12]		
79/M	FNA	Failure	Undifferentiated pleomorphic sarcoma	-
	ST	46,XY[21]		
46/F	FNA	46,XX[7]	Undifferentiated pleomorphic sarcoma	
	ST	82-92,-X,-X,-X,-X,del(1)(q12),del(1)(q21),der(2)del(2)(p12)t(1;2)(q21;q33)ins(2;?)(q33;?),del(3)(p12),del(5)(p12),add(6)(q13)x2,der(7)add(7)(p22)add(7)(q32),add(9)(q34),del(10)(p13),add(12)(p13),add(12)(q24),del(12)(p12),add(13)(p13),add(16)(q13),der(16)t(1;16)(q12;q12),del(17)(p11),add(19)(q13),add(20)(q13),inc[cp8]		7478:251
74/F	FNA	Failure	Undifferentiated pleomorphic sarcoma	-
	ST	46,XX[10]		
68/M	FNA	**72-73,XXY,?add(12)(q13),+mar,inc**[6]/46,XY[8]	Undifferentiated pleomorphic sarcoma	-
	ST	74-80,XXY,+2,+4,+7,+7,-8,-10,+der(12)t(?3;12)(q21;q13),+add(12)(q15),+13,-16,add(18)(p11),+19,-22,+2-3mar[6]/46,XY[8]		
57/M	FNA	Failure	Undifferentiated pleomorphic sarcoma	-
	ST	63-73,X?,del(1)(q32),add(7)(p22),del(10)(p11),der(?12)add(12)(p11)add(12)(q24),add(19)(q13),+der(?)t(?;3)(?;p11),+der(?)t(?;5)(?;q1?1),+3-4mar,inc[cp5]/46,XY[3]		

Bone tumors

Cartilage tumors

19/M	FNA	Failure	Chondroma	-
	ST	46,XY[24]		
69/M	FNA	Failure	Chondrosarcoma	
	ST	75-79,+X,add(X)(q27)x2,-Y,der(1)t(1;2)(q32;q21),add(3)(q27),+der(3)t(3;14)(q21;q11)ins(3;?)(q21;?),-4,+5,+5,+5,+6,+7,+7,+7,-8,-9,-9,-10,der(10)t(5;10)(q21;p15)x2,add(11)(q25),+12,+12,+12,-13,der(13)t(2;13)(p11;p11),-14,add(14)(p?),-15,-15,+16,+17,add(17)(p13)x2,+18,+19,+19,add(19)(q13)x3,+20,add(20)(p13)x2,der(20)t(10;20)(q11;p13)x2,-21,-21,-21,inc[cp20]/46,XY[2]		9347:42
51/M	FNA	Failure	Chondrosarcoma	-
	ST	46,XY[19]		
52/M	FNA	46,XY[24]	Chondrosarcoma	
	ST	38,X,-Y,add(1)(p21),-4,-6,der(8)t(8;?22)(p12;q11),-9,-10,-11,add(11)(p15),-14,der(17;21)(q10;q10),add(18)(p11),add(19)(p13),-22,der(22)t(?9;22)(q13;p11),+mar[cp6]		12869:32

Osteogenic tumors

23/M	FNA	46,XY[4]	Osteoblastoma	-
	ST	44,X,-Y,add(1)(p34),del(1)(q21),del(2)(p21p23),+3,del(3)(p12)x2,del(6)(q15),der(6)t(6;13)(q27;q12),+10,der(10)t(8;10)(q11;q26)x2,del(12)(p11),-13,+der(15)t(1;15)(q21;p13)ins(1;?)(q32;?),+16,-17,add(17)(q11),-18,-22,-22[15]/43,idem,-15[3]/46,XY[3]		
15/F	FNA	Failure	Osteosarcoma	
	ST	75-89,XX,-X,add(1)(q11)x2,add(1)(q21),+add(1)(q21),+del(1)(q21q25)x2,add(2)(p16)x2,del(3)(p11),del(6)(q?),-9,inc[34]/47,XX,+marc[14]		5007:11
17/M	FNA	Failure	Osteosarcoma	
	ST	77-83,X,-X,-Y,-1,+2,add(2)(p12)x2,-3,+5,+6,+8,+8,der(8)t(3;8)(p23;p21)x3,+11,+12,-13,-13,-13,+14,+17,der(17)t(1;17)(p13;p11)x2,-18,-19,add(19)(p13)x2,+20,+add(20)(p13),+7-9mar[20]		5007:19
15/M	FNA	**56,XY,del(1)(q11),del(2)(p21),add(12)(p13),+der(?)t(?;13)(?;q14),inc**[3]/46,XY[8]	Osteosarcoma	5007:12
	ST	54-57,XY,del(1)(q11),del(1)(q32),der(1)add(1)(p11)del(1)(q42),del(2)(p21),add(4)(p11),der(20)t(7;20)(q21;q13),+der(?)t(?;5)(?;q13)hsr(?;5)(?;q13),+der(?)t(?;13)(?;q14),inc[cp7]		5007:12
16/M	FNA	**56-60,XXY,inc[4]/104-117,XX,add(1)(q21),del(2)(p12),del(6)(q16),+der(?)t(?;2)**	Osteosarcoma	5007:15

		(?;q13),inc[6]/46,XY[5]		
	ST	46,XY[19]		
10/F	FNA	Failure	Osteosarcoma	-
	ST	46,XX[7]		
56/F	FNA	**43-48,X,-X,ins(1;?)(q21;?),+add(3)(q11),-4,del(5)(p12),add(6)(p25),del(6)(p23),** **+add(7)(p22),-8,-9,add(10)(q22),-11,add(11)(p15)x2,-13,der(13;?)t(13;?)(q34;?)** **t(?;13)(?;q12),-14,-15,der(18)add(18)(p11)hsr(18)(p11),+4mar**[cp8]/46,XX[2]	Osteosarcoma	-
	ST	46-49,X,-X,ins(1;?)(q21;?),add(3)(q11),-4,del(5)(p12),add(6)(p25),add(7)(p22),-8,-9, -10,add(11)(p15)x2,-13,der(13;?)t(13;?)(q34;?)t(?;13)(?;q12),-14,-15,der(18) add(18)(p11)hsr(18)(p11),+4mar,inc[cp6]/46,XX[2]		
80/M	FNA	Failure	Osteosarcoma	-
	ST	77-80,X?,add(1)(p36),inc[2]/46,XY[22]		
22/F	FNA	Failure	Osteosarcoma	-
	ST	46,XX[25]		
19/M	FNA	46,XY[13]	Osteosarcoma	-
	ST	46,XY[13]		
32/F	FNA	Failure	Undifferentiated pleomorphic sarcoma	8379:1517

	ST	46-47,X,?del(X)(q?),add(1)(q21),add(3)(p11),add(3)(q12),add(4)(p1?),?add(7)(q22),add(9)(p11),+2mar,inc [cp5]		

Ewing sarcoma/primitive neuroectodermal tumor

18/M	FNA	**46,Y,t(X;4)(p11;q31)**[25]	Primitive round cell sarcoma	11395:1
	ST	46,Y,t(X;4)(p11;q31)[28]/46,XY[15]		11395:1
27/M	FNA	46,XY[14]	Ewing sarcoma	-
	ST	49-52,XY,+Y,+4,+5,+7,+8,+10,t(11;22)(q24;q12)[cp8]/46,XY[33]		
17/M	FNA	**55,X,-Y,+der,(Y)t(Y;1)(q12,q21),+der(Y)t(Y;1),+5,+6,+8,+8,+9,t(11;22)(q24;q12),+12,-15,+der(15)t(2;15)(p11;p13),+17,+20**[14]	Ewing sarcoma	12093:16
	ST	46,XY[12]		
10/M	FNA	**46,XY,t(15;19)(q14;p13)**[8]/**45,X,-Y,t(15;19)(q14;p13)**[14]	Ewing sarcoma	12820:1
	ST	45,X,-Y,t(15;19)(q14;p13)[cp4]		12820:1
27/M	FNA	46,XY[6]	Ewing sarcoma	-
	ST	46,XY[25]		
	FNA[d]	**69-70,XY,-X,+Y,del(1)(p22),+8,-9,der(11)?inv(11)(p14q12)t(11;22)(q?;q?),+13,-14,-15,add(16)(q2?2),-17,+?19,+20,+20,add(21)(q22),-22,der(22)t(11;22)**[19]		
22/M	FNA	**47-49,XY,add(22)(q12),inc**[2]	Ewing sarcoma	12093:21

	ST	46,XY[32]		
15/M	FNA	Failure	Ewing sarcoma	-
	ST	46,XY[31]		
12/F	FNA	46,XX[3]/*46,XX,t(1;11;22)(q23;q24;q12)[1]*	Ewing sarcoma	-
	ST	46,XX,t(1;11;22)(q23;q24;q12)[3]		
15/F	FNA	46,XX[13]	Ewing sarcoma	-
	ST	46,XX,?del(22)(q12)[5]/46,XX[4]		
8/F	FNA	**46,XX,t(11;22)(q24;q12)**[8]	Ewing sarcoma	-
Miscellaneous bone tumors				
38/M	FNA	46,XY[6]	Giant cell tumor	
	ST	45-46,XY,tas(8;17)(p23;p13)[cp4]/45-46,XY,tas(8;19)(p23;p13)[cp3]/45-46,XY, tas(7;17)(q36;p13)[cp2]/46,XY[39]		12830:53
31/F	FNA	Failure	Giant cell tumor	
	ST	45-46,XX,inv(3)(p13q21)[cp12]/46,XX,t(12;13)(q15;p11)[10]/46,XX,t(4;11) (q21;p15)[6]/46,XX,t(2;14)(q33;q32)[5]/46,XX,t(2;11)(q21;q25)[4]/46,XX,t(2;15) (q23;q22)[4]/46,XX,t(11;19)(q21;p13)[4]/46,XX,t(2;11)(p23;q13)[3]/46,XX,t(4;13) (q21;q34)[3]/46,X,del(X)(q24),t(10;13)(q11;q14)[2]/46,XX,tas(1;6)(q44;q27)[2]/		12830:55

		46,XX,t(1;9)(q32;q32),t(5;12)(q11;p11)[2]/46,XX,t(2;9)(q37;q22)[2]/46,XX,t(3;10)(q12;q26),inv(8)(p21q24)[2]/46,XX,t(7;15)(p15;q24)[2]/46,XX,t(9;19)(q22;q13)[2]/46,XX,t(10;21)(p11;q22)[2]/46,XX[175]		
48/F	FNA	Failure	Giant cell tumor	
	ST	46,XX,t(5;8)(q35;q22) [4]/46,XX[21]		12830:66
74/M	FNA	Failure	Chordoma	
	ST	40-42,XY,-3,der(6)t(6;9)(q?25-27;q11-12),-8,-9,der(9)t(9;10)(p24;?) or der(9)t(9;16)(p24;?),-10,dic(12;?16)(?p1?3;?)?inv(12)(p11p13),der(21)t(8;21)(q11;p13),-22[5]/46,XY[5]		10597:1
41/M	FNA	46,XY[29]	Chordoma	-
	ST	46,XY[22]		

[a] FNA = fine needle aspiration biopsy; ST = surgical tumor biopsy. An asterisk indicates that a core needle biopsy was analyzed cytogenetically from the same case (see Table 2).

[b] Clonal, acquired chromosome aberrations in FNA biopsies are indicated in bold. Italics indicate a likely tumor associated aberration found in a single cell (hence not clonal).

[c] Previously published karyotypes are referred to with reference number and case number in Mitelman Database of Chromosome Aberrations in Cancer 2010.

[d] ST and FNA were from different lesions, e.g., primary tumor and local recurrence.

Table 2. Cytogenetic findings in cell cultures from core needle aspiration biopsies and corresponding surgical tumor biopsies. Clinical data and karyotypes in 30 patients with soft tissue or bone tumors.

Age/sex	Type of sample[a]	Karyotype[b]	Diagnosis	Reference[c]
		Soft tissue tumors		
Adipocytic tumors				
78/M	CN*	**45,XY,der(2)t(2;?13)(q13-21;q21-31),add(13)(q13),-16**[11]	Spindle cell lipoma	-
	ST	45,XY,der(2)t(2;?13)(q13-21;q21-31),add(13)(q13),-16[16]		
62/F	CN	Failure	Atypical lipomatous tumour	-
	ST	43-50,XX,add(4)(q3?),-6,+1-2r,+mar[cp7]/46,XX[4]		
65/M	CN	**46,XY,del(1)(p31)**[22]	Atypical lipomatous tumour	-
	ST	46,XY,del(1)(p31)[24]		
53/F	CN	46,XX[22]	Atypical lipomatous tumour	-
	ST	47,XX,+r[8]/47,X,-X,+?12,+r[10]		
60/M	ST*	46,XY,t(12;13;16)(q13;q12;p11)[21]/46,XY[4]	Myxoid liposarcoma	12980:72
	CN[d]	**46,XY,t(2;18)(q35;q11),t(12;13;16)(q13;q12;p11)**[10]		

	CN[d]	**46,XY,t(2;18)(q35;q11),t(12;13;16)(q13;q12;p11)**[3]		
30/F	CN	**46,XX,t(12;16)(q13;p11)**[7]	Myxoid liposarcoma	-
	ST	46,XX,t(12;16)(q13;p11)[11]		
57/F	CN	Failure	Myxoid liposarcoma	-
	ST	46,XX[6]		
81/M	CN	**45,X,-Y**[3]**/**46,XY[12]	Pleomorphic liposarcoma	-
	ST	67-73,X?,inc[cp16]/120-130,X?,inc[cp5]/46,XY[18]		
Fibroblastic/Myofibroblastic tumors				
52/F	CN	46,XX[16]	Desmoid type fibromatosis	-
	ST	46,XX[35]		
44/F	CN	46,XX[11]	Desmoid type fibromatosis	-
	ST	46,XX[23]		
50/F	CN	Failure	Desmoid type fibromatosis	-
	ST	46,XX[25]		
64/F	CN	46,XX[4]	Myxofibrosarcoma	-
	ST	43,X?,del(1)(q21),?der(12)hsr(12)(q13)add(12)(q24),+?r,inc[cp4]		
56/F	CN	46,XX[20]	Myxofibrosarcoma	-

	ST	46,XX[11]		
31/M	CN	46,XY[13]	Myxofibrosarcoma	-
	ST	48-52,X,-Y,+7,+8,?-9,der(14;14)(q10;q10),-16,+17,+18,+2mar,inc[cp6]/46,XY[2]		
25/M	CN	Failure	Low grade fibromyxoid sarcoma	-
	ST	46,XY[12]		
80/M[e]	CN	**46,XY,t(9;17)(q34;q23),?t(10;14)(p14;q24)[3]/46,XY,t(11;12)(q13;q24)[3]**	Myxofibrosarcoma	-
	ST	51-59,XY,add(1)(q21)x2,-4,-4,-6,add(6)(q15),?der(6)del(6)(p23),+7,-9,add(10)(p1?), ?der(11)add(11)(p13)del(11)(q23),del(12)(q24),add(13)(p11),?i(14)(q10), add(15)(p11),?add(16)(q22),-17,-18,-18,inc[cp7]/95-109,idemx2[cp2]		
Smooth muscle tumors				
61/F	CN	46,XX[25]	Leiomyoma	-
	ST	44,XX,add(1)(p11),del(6)(q11),-10,-14[12]/86-87,idemx2[3]		
69/M	CN	Failure	Leiomyosarcoma	-
	ST	Failure		
Soft tissue tumors of miscellaneous/uncertain differentiation				
36/M	CN	**46,XY,t(2;18)(q3?3;q23),del(3)(p13p23)[11]**	Neurofibroma	
	ST	46,XY[7]		

47/F	CN	**45,X,t(X;18)(p11;q11),der(1)t(1;3)(q42;q11),-3[7]/46,idem,+mar[6]**	Synovial sarcoma	-
	ST	46,X,t(X;18)(p11;q11)[3]/45,idem,der(1)t(1;3)(q42;q11),-3[5]/46,idem, der(1)t(1;3)(q42;q11),-3,+mar[4]		
24/M	CN	**40-41,Y,t(X;18)(p11;q11),-3,?inv(4)(p15q24),-9,-11,-15,-17,-20**[cp3]**/58-59,XY, t(X;18)(p11;q11),-1,-3,-4,-7,-10,-18,-19,-21,-22**[cp2]/46,XY[5]	Synovial sarcoma	-
	ST	41,Y,t(X;18)(p11;q11),-3,?inv(4)(p15q24),-9,-11,-15,-20[9]/46,XY[6]		
36/M	CN	46,XY[21]	Undifferentiated pleomorphic sarcoma	-
	ST	46,XY[25]		
66/M	CN	46,XY[11]	Undifferentiated pleomorphic sarcoma	-
	ST	Failure		
86/M	CN	**60-70,X?,inc**[11]/46,XY[5]	Undifferentiated pleomorphic sarcoma	-
	ST	Failure		

Bone tumors

Cartilage tumors

23/F	CN	46,XX[17]	Chondrosarcoma	-
	ST	46,XX[24]		

Osteogenic tumors

62/M	CN	**46,Y,?t(X;11)(p21;q13),add(3)(?p21),-5,-10,der(20)t(5;20)(q11;q13)add(5)(q3?5), +2mar**[9]	Osteosarcoma	-
	ST	46,Y,t(X;11)(p21;q12),add(3)(p?21),-5,del(6)(q15),-10,der(20)t(5;20)(q11;q13) ins(5;?)(q3?1;?),+r,+mar[14]/46,XY[3]		
18/M	CN	Failure	Osteosarcoma	-
	ST	Failure		
16/M	CN	**32-38,XY,add(1)(q42),der(1)t(1;9)(q32;q13)ins(1;?)(q32;?),del(2)(q33),-3,der(3) t(3;15)(q29;q13)ins(3;?)(q29;?),add(4)(q35),der(6)t(6;7)(q15;q22)ins(6;?)(q15;?), +add(6)(q1?5),del(7)(q11),del(8)(p21),der(8)t(?1;8)(q32;q22)ins(8;?)(q22;?),-9, -10,-10,-11,add(12)(q13),-13,-14,-15,-16,i(16)(q10),-17,-17,-19,ins(19;11) (q13;q13q25),-20,add(20)(p13),-22,+3mar[cp13]**	Osteosarcoma	-
	ST	35-38,XY,add(1)(q42),der(1)t(1;9)(q32;q13),ins(1;?)(q32;?),del(2)(q33),-3,der(3) t(3;15)(q29;q13)ins(3;?)(q29;?),der(6)t(6;7)(q15;q22)ins(6;?)(q15;?),+add(6)(q1?5), del(8)(p21),der(8)t(?1;8)(q32;q22)ins(8;?)(q22;?),-9,-10,-10,-11,add(12)(q13),-13, -14,-15,-16,-17,-17,-18,-19,ins(19;11)(q13;q13q25),-20,add(20)(p13),-21,-22, +4mar[cp10]		

Ewing sarcoma/primitive neuroectodermal tumor

21/M	CN	**36-38,XY,-3,-5,-6,+8,-9,-11,-14,-15,add(16)(p11),-17,-19,-22,der(22)t(11;22)(q24;q12)[cp6]/54-55,XY,+X,+2,+5,+8,+8,t(11;22)(q24;q12),+12,+20,+21[cp3]**	Ewing sarcoma	-
	ST	Failure		
Giant cell tumor				
25/M	CN	**47,XY,+11**[11]/46,XY[6]	Giant cell tumour	-
	ST	47,XY,+11[4]/46,XY[19]		

[a] CN = core needle aspiration biopsy; ST = surgical tumor biopsy. An asterisk indicates that a fine needle biopsy was analyzed cytogenetically from the same case (see Table 1).

[b] Clonal, acquired chromosome aberrations in FNA biopsies are indicated in bold.

[c] Previously published karyotypes are referred to with reference number and case number in Mitelman Database of Chromosome Aberrations in Cancer 2010[5].

[d] ST and CN were from different lesions, e.g., primary tumor and local recurrence.

[e] Patient who had received prior radiotherapy.

Table 3. Cytogenetic findings in soft tissue tumors

Karyotype	Fine needle aspirations (FNA)	Core needle biopsy (CN)	Surgical tumor biopsy (ST)
Failure	46 (55%)	5 (20%)	6 (6%)
Normal	19 (23%)	9 (36%)	30 (29%)
Abnormal	18 (22%)	11 (44%)	67 (65%)
Total	83	25	103

Table 4. Cytogenetic findings in bone tumors

Karyotype	Fine needle aspiration (FNA)	Core needle biopsy (CN)	Surgical tumor biopsy (ST)
Failure	13 (42%)	1 (17%)	2 (6%)
Normal	9 (29%)	1 (17%)	12 (34%)
Abnormal	9 (29%)	4 (67%)	21 (60%)
Total	31	6	35

Table 5. Cytogenetic findings in benign and malignant tumors[a]

	FNA			CN			ST		
	Failure	Normal	Abnormal	Failure	Normal	Abnormal	Failure	Normal	Abnormal
Benign tumor	19 (54%)	11 (31%)	5 (14%)	2 (20%)	4 (40%)	4 (40%)	0	16 (36%)	29 (64%)
Malignant tumor	40 (51%)	17 (22%)	22 (28%)	4 (19%)	6 (29%)	11 (52%)	8 (9%)	26 (28%)	59 (63%)
Total	59 (52%)	28 (25%)	27 (24%)	6 (19%)	10 (32%)	15 (48%)	8 (6%)	42 (30%)	88 (64%)

[a] FNA=Fine needle aspiration, CN=Core needle biopsy, ST=Surgical tumor biopsy

Table 6. Cytogenetic outcome depending on type of tumor[a]

	FNA			CN			ST		
	Failure	Normal	Abnormal	Failure	Normal	Abnormal	Failure	Normal	Abnormal
Adipocytic tumors	15 (65%)	4 (17%)	4 (17%)	2 (22%)	1 (11%)	6 (67%)	0	5 (17%)	24 (83%)
Fibroblastic/Myofibroblastic tumors	12 (52%)	5 (22%)	6 (26%)	2 (25%)	5 (63%)	1 (12%)	1 (3%)	9 (30%)	20 (67%)
Smooth muscle tumors	8 (50%)	4 (25%)	4 (25%)	1 (50%)	1 (50%)	0	2 (12%)	1 (6%)	14 (82%)
Osteogenic tumors	6 (55%)	2 (18%)	3 (27%)	1 (33%)	0	2 (67%)	1 (8%)	4 (31%)	8 (62%)
Ewing sarcoma/primitive neuroectodermal tumour	1 (9%)	3 (27%)	7 (64%)	0	0	1 (100%)	1 (10%)	4 (40%)	5 (50%)

[a] FNA= Fine needle aspiration, CN=Core needle biopsy, ST=Surgical tumor biopsy