



LUND UNIVERSITY

Chromosome banding analysis of cells from fine-needle aspiration biopsy samples from soft tissue and bone tumors: is it clinically meaningful?

Walther, Charles; Domanski, Henryk; Vult von Steyern, Fredrik; Mandahl, Nils; Mertens, Fredrik

Published in:
Cancer Genetics

DOI:
[10.1016/j.cancergen.2011.01.001](https://doi.org/10.1016/j.cancergen.2011.01.001)

2011

[Link to publication](#)

Citation for published version (APA):

Walther, C., Domanski, H., Vult von Steyern, F., Mandahl, N., & Mertens, F. (2011). Chromosome banding analysis of cells from fine-needle aspiration biopsy samples from soft tissue and bone tumors: is it clinically meaningful? *Cancer Genetics*, 204(4), 203-206. <https://doi.org/10.1016/j.cancergen.2011.01.001>

Total number of authors:
5

General rights

Unless other specific re-use rights are stated the following general rights apply:
Copyright and moral rights for the publications made accessible in the public portal are retained by the authors and/or other copyright owners and it is a condition of accessing publications that users recognise and abide by the legal requirements associated with these rights.

- Users may download and print one copy of any publication from the public portal for the purpose of private study or research.
- You may not further distribute the material or use it for any profit-making activity or commercial gain
- You may freely distribute the URL identifying the publication in the public portal

Read more about Creative commons licenses: <https://creativecommons.org/licenses/>

Take down policy

If you believe that this document breaches copyright please contact us providing details, and we will remove access to the work immediately and investigate your claim.

LUND UNIVERSITY

PO Box 117
221 00 Lund
+46 46-222 00 00

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

Charles Walther^{a, b, *}, Henryk A. Domanski^a, Fredrik Vult von Steyern^c, Nils Mandahl^b, Fredrik Mertens^b

^aDepartment of Pathology, University and Regional Laboratories, Lund University Hospital, Lund University, Lund, Sweden

^bDepartment of Clinical Genetics, University and Regional Laboratories, Lund University Hospital, Lund University, Lund, Sweden

^cDepartment of Orthopedics, Clinical Sciences, Lund, Lund University and Skane University Hospital, Sweden

*Corresponding author: Dr. Charles Walther, Department of Pathology, University and Regional Laboratories, Lund University Hospital, Lund University, SE-221 85 Lund, Sweden. Tel:+4646173510; Fax+4646143307; E-mail Charles.Walther@med.lu.se

Supported by: the Swedish Childhood Cancer Foundation and the Lund University Hospital Funds.

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.

Acknowledgments

This study was supported by the Swedish Childhood Cancer Foundation and the Lund University Hospital Funds.

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 literature. *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]/ 46,idem,tas(Y;21)(p11;p13)[3] | | 9986:35 |
| 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

