Supplementary Table 1 List of publications reporting individuals with Jaffe-Campanacci syndrome

NO.	Ref.	Age (years)	Gender	Race/Ethnicity	Non-ossifying Fibroma	Other Bony Abnormalities	CALS	Neurofibroma	Axillary Freckling	Mental retardation	Cardiovscular malformations	Scoliosis	Cerebral/neurologic abnormalities	Ocular anomalies Lisch Nodules	Alopecia	Short stature	Growth hormone deficiency	Precocious puberty	Other manifestations	Past illnesses	Family History	Fulfill NIH Criteria for Diagnosis of NF1	Clinical Diagnosis	Genetic Testing	Treatment for JCS	Prognosis
1	Mirra et al., 1982	14	Female	White	Distal of both femurs, proximal of both tibias and fibula, distal of left tibia	Mandible with multiple lytic foci consistent with giant cell reparative granulomas	Numerous	None	On entire body, especially around lips	None	None	None	None	Eye exam done, no comment on Lisch nodules	None	None	None	None	None	Amoebic dysentery, pneumonia and hepatitis	No history of NF1	NA	JCS, but NF1, others considered	NA	Curettage	NA
2	Campanacci et al., 1983	11	Male	ND	Mainly right sided	Four pathologic fractures, vagus deformity of the knee	Had, but not specified	None	None	Had, but not specified	Had, but not specified	None	None	None	None	None	None	None	None	None	None	NA	Proposed multiple NOF plus other extraskeletal congential anomalies may constitute a new syndrome	NA	Routine treatmen for fracture	Healed in the normal time.
3	Campanacci et al., 1983	17	Male	ND	Lower limbs	None	Had, but not specified	None	None	Had, but not specified	Had, but not specified	None	None	None	None	None	None	None	None	None	None	NA	Proposed multiple NOF plus other extraskeletal congential anomalies may constitute a new syndrome	NA	NA	NA

4	Campanacci et al., 1983	11	Male	ND	Bilateral symmetrical ,involving the humerus, radius, ilium, femur, tibia and fibula	Two pathological fractures, Valgus deformity of the knee	Right side of the trunk, right arm and right leg	None	None	Had, but not specified	None	None	None	Bilateral conjunctival dermolipoma, right corneal leucoma, evidence of tarsorrhaphy of the lateral third of the eye	Alopecia,	None	None	None	None	None	No relevant family history	NA	Proposed multiple NOF plus other extraskeletal congential anomalies may constitute a new syndrome	NA	Routine treatmen for fracture	Healed in the normal time.
5	Campanacci et al., 1983	12	Male	ND	Lower limbs	None	Had, but not specified	None	None	Had, but not specified	None	None	None	None	None	None	None	Precocious puberty	None	None	None	NA	Proposed multiple NOF plus other extraskeletal congential anomalies may constitute a new syndrome	NA	NA	NA
6	Campanacci et al., 1983	14	Male	ND	Diffuse	One pathological fractures, Mega-ureter	Right sided	None	None	None	None	None	None	Had, but not specified	None	None	None	None	None	None	None	NA	Proposed multiple NOF plus other extraskeletal congential anomalies may constitute a new syndrome	NA	Routine treatmen for fracture	Healed in the normal time.

7	Campanacci et al., 1983	6	Male	ND	Right lower limb	One pathological fractures, Valgus deformity of the knee	Had, but not specified	None	None	None	None	None	None	None	None	None	None	None	None	None	None	NA	Proposed multiple NOF plus other extraskeletal congential anomalies may constitute a new syndrome	NA	Routine treatmen for fracture	Healed in the normal time.
8	Steinmetz et al., 1988	15	Male	White	Distal femora, proximal tibia	Pathologic fracture, right distal femur	"Multiple"	None	None	None	None	None	None	None	None	None	None	None	None	None	None	NA	NF1 then JCS	NA		
9	Blau et al., 1988	4	Female	White	Proximal and distal of femer and tibia, distal left fibula	7 pathologic fractures after minor trauma; "Dramatic" local growth of NOF	Left axilla, the left scapular region, and the posterior aspect of the left upper arm	None	Multiple "light-brown macules" in left axilla	None	None	None	None	None	None	None	None	None	None	None	None	NA	Multiple NOF, presumably JCS	NA	All pathologiacl fracturs were treated in a long cast; three years later,all femoral and tibial lesions were curetted, bone-grafting was done using homologous banked bone. and both femora and tibiae were fixed internally using the Bailey- Dubow elongating-rod system.	The pathological fractures united in the expected length; Three years later,right genu valgum measuring 20 degrees and a limb-length discrepancy of 2.5 centimeters.Fifteen months later, the left ankle lacked I5 degrees of dorsiflexion and the right ankle lacked 10 degrees. The right lower extremity was clinically 3.5 centimeters shorter than the left.

10	Kotzot et al., 1994	27	Female	White	Left proximal and distal tibia, left distal and proximal femur, left 3rd and 5th ribs	None	Left sided only multiple large CALS	None	None	None	Stenosis of the aortic isthmus	Moderate scoliosis	None	None	None	None	None	None	Chylothorax,Chylopericardium	None	None	NA	JCS	None	The lesiaons of left proximal and distal tibia, left distal and proximal femur were cureted at 12 to 16 years.	The left leg was 6cm than the right.
11	Hau et al., 2002	15	Male	ND	Distal femora, proximal left tibia and fibula, proximal right tibia	Pathologic fracture in left distal femur	Variously sized, smooth-margined “coast of California” café-au-lait spots on the trunk	None	Prominent bilateral axillary	Mildly to moderately retarded	None	None	None	None	None	None	None	None	None	None	None	None	NA	NF1, then JCS	Not specified	Two previous surgical procedures consisting of curettage and bone graft packing for lesions in the distal parts of both femora two years before admission. The patient underwent intralesional excision, curettage, allograft strut-grafting, and plate fixation of the left femoral lesion. Six months later, underwent a similar operation on the right femur.	One year later, both treated lesions had healed and the grafts were well incorporated without signs of recurrence.

12	Colby et al., 2003	13	Female	ND	Bilateral distal femur, tibia and fibula	Pathologic fracture in left femur	Multiple large CALS over the limbs and torso	None	Axillary	Had trouble in school and typical garades included Cs, Ds and Fs	None	None	None	No Lisch, but an medial ectopic pupil of the left eye	None	None	None	None	None	None	No features of NF1 and bone lesions	Diagnosed based on CALS and Axillary Freckling	NF1, then JCS	Not specified	Routine treatmen for fracture	Fracture healed
13	Colby et al., 2003	15	Male	ND	Bilateral femurs, tibias and fibulas	Pathologic fracture in left femur	Multiple truncal CALS	None	Axllary and inguinal freckling	IQ(WISC-III) was 82	None	None	Cranial lmaging showed enhanced signal density in the brain stem, basal ganglia, thalamus, cerebellum and white matter; Mild hydrocephalus	Lisch nodules were present	None	None	None	None	None	Attention deficit disorder	No features of NF1 and bone lesions	Diagnosed based on CALS and axillary freckling	NF1, then JCS	Not specified	Surgical fixation of the NOFs and fracture with plate and screws for left femur; Bone graft in the right femur	ND
14	Colby et al., 2003	13	Male	ND	Right distal femur, right proximal tibia and left proximal tibia	ND	On the torso	On the torso	None	Psychometric testing: low normal - mild mental retardation	None	None	MR scan: several signal variations; Mild balance and gait abnormalities	Palpebral fissures, mild ptosis and Lisch nodules	None	None	None	None	None	None	No features of NF1 and bone lesions	Had, but not specified	NF1, then JCS	Not specified	Surgeries on right distal femur, right proximal tibia and left proximal tibia	ND

15	Colby et al., 2003	17	Male	ND	Bilateral tibias and fibulas	Pseudoarthrosis of the right lower leg; Velopharyngeal insufficiency	Multiple	On the torso	Axllary and inguinal freckling	IQ was 74	None	Mild scoliosis	Mild unsteady gait	Mildly down slanting palpebral fissures and Lisch nodules	None	None	None	None	None	None	No features of NF1 and bone lesions	Diagnosed based on CALS and pseudoarthrosis of the right lower leg	NF1, then JCS	Molecular testing of the NF1gene showed a partial deletion	ND	ND
16	Nezhad et al., 2003	7	Male	ND	Proximal and diatal of the right tibia, femur and right humerus diaphysis	Pathologic fracture in right tibia; The skull was abnormal shape	Multiple "Coast of California" CALS on the right side of torso	None	None	None	Mild cardiomegaly, Atrial septal defect and coarction of aorta	None	None	Right eye amblyopia with sceroma and optic nerve hypoplasia	Alopecia on the right occiput	None	None	None	None	None	None	Negative for neurofibromatosis or any related disorders	ND	JCS	ND	Intralesional excision, curettage and allograft strut grafting of the proximal tibial lesion	ND
17	Al-Rikabi et al., 2005	6	Male	ND	Right upper tibia bone lesion"; similar lesions observed radiographically in right distal femur a distal tibia	Right severe genu varus deformity with leg shortening	Large confluentCALS from pubis to right thigh	None	None	None	None	None	None	None	None	None	None	None	None	None	None	ND	Diagnosed based on CALS	NF1 then JCS	Not specified	Curettage and bone grafting of the right upper tibia	ND

18	Kourelis et al., 2012	21	Female	ND	Multiple old NOF in left humerus, pelvis	Multiple pathologic fractures in the past; A small pulmonary embolus in a subsegmental branch	Numerous	None	Axillary	None	None	None	None	None	None	None	None	None	None	None	None	Unknown - adopted	"…patient met 3 of the … criteria for the diagnosis of NF1… more thn 6 CALM, axillary freckling and thinning of the long bone cortex."	JCS	Not specified	ND	ND
19	Sonar et al., 2012	13	Male	White	Distal left tibia and femur	Pathologic fracture of distal part of left tibia	"Variously sized café-au-lait spots," location not specified	None	Axillary	IQ was 69	None	None	None	None	None	None	None	None	None	None	Cryptorchidism	None of the family members had skin lesions, such as café-au-lait spots, axillary freckling and long bone fracture history. Family research showed no genetic errors	ND	"Neurofibromatosis" then JCS	ND	Curettage, grafting with allograft, and fixation with conventional plate of the left tibia and femur	Both tibial and femoral fractures healed on time; No signs of recurrence

20	Yang et al., 2012	2	Female	Yellow	Proximal of the right humerus, distal of the right radius and femur, and femoral shaft	Pathologic fracture of the right humerus and femur	Multiple large ""coast of Maine" CALS over the right face and neck, right limbs, and right torso	None	None	None	None	None	None	None	None	None	None	None	None	None	Negative for features of NF1 and bone lesions	Dignosed, but not specified	NF1, then JCS	ND	Curettage and bone grafting of the right humerus and femur	3 months later, the pain relieved and normal function in both the upper and lower right limbs
21	Cherix et al., 2014	17	Female	ND	Bilateral distal femurs and proximal tibias	Pathologic fracture of the right distal femoral diaphysis	Multiple "coast of California" CALS on the abdomen, the back and all four limbs	None	Bilateral axillary freckles	Mild mental retardation	None	None	None	A Lisch nodule	None	None	None	None	None	None	Her father suffered from NF1	Dignosed, but not specified	NF1, then JCS	ND	Open recuction and internal plate fixation of the right distal femoral diaphysis; the others performed curettage and bone grafting	The fracturs healed without complications, and the patient returned to work witnin 4 months; the others healed after surgery
22	Eun MC at al, 2016	9	Female	Yellow	The left proximal humerus, left distal femur,both proximal tibias, and left proximal fibula		Multiple CALS on the left side of her face, trunk, and left extremity	None	None	She displayed 2 years of retarded development	A grade I–II systolic heart murmur was heard on auscultation of the chest; aortic coarctation	None	Unilateral ventricular dilatation and an extra-axial cerebrospinal fluid space in the left hemisphere and posterior fossa were observed	Incomplete upper eyelids (coloboma with ectopic cilia on the left side and cryptophthalmos with dermolipoma on the right	Sparse hair with separated alopecia	Her height was 110 cm, which was under the 3rd percentile for her age	Growth hormone (GH) level was below the normal range. Using the insulin stimulation and levo-dihydroxyphenylalanine tests, GH deficiency was confirmed	None	Edema of the left lower extremity	None	None	None	Encephalocraniocutaneous lipomatosis, JCS	None	ND	ND

Diagnositic criteria for neurofibromatosis type 1 (NF1) was published in 1988. The *NF1* gene was identified in 1990. NA: Not applicable; ND: No data; NOF: Non-ossifying fibroma; CALM: Café-au-lait macule.