

Supplementary Table 1: Description of the patients affected with Ollier disease\*. French cohort.

		PTHR1 screening		PTHR1 variant tissue / blood	Sexe	Age at Dg <sup>1</sup> (year)	Clinical data	
		blood	tissue				enchondroma s	enchond studied
1	B371	Yes	/		F	1,08	femur tibia right	
2	B285	Yes	/	p.T163Tp. E546K (na /+)	F	1,2	hand right	
3	HP3	Yes	/		F	2	hand, arm, leg right + left	
4	HP22	Yes	/		F	2	hand foot right + left	
5	HP26	Yes	/		F	2	femur left	
6	HP42 HP78	Yes	Yes		F	2	arm, leg right + left	hand R <sup>1</sup> (EC)
7	HP74	Yes	Yes		F	1	femur right	femur R
8	HP65	Yes	/		F	2	na	
9	HP60	Yes	/		F	2,06	hand humerus foot femur left	
10	HP25 HP100	Yes	Yes		F	3	hand left and right, shoulder left, pelvis, toe right	humerus L (EC)
11	HP37	Yes	/	p.A72A (na /+)	F	3	hand right, humerus left, toes left	
12	HP66 HP54	Yes	Yes	p.R255H (+ / +)	F	3	hand foot right + left	hand phalangis digit 1,4,5 R (EC)
13	HP29 HP80	Yes	Yes		F	4	femur tibia fibula left	tibia L (EC)
14	HP41	Yes	/		F	4	hands left + right, foot right	
15	HP12	Yes	/		F	4	na	
16	HP11	/	/		F	5	pelvis, femur, ankle right	
17	HP7	Yes	/		F	7	femur right	
18	HP23 HP95	Yes	Yes		F	7	hand cubitus left	hand phalangis digit 2-3 (EC)
19	HP68 HP27	Yes	Yes		F	7	wrist left + right, humerus left, femur left	femur L (EC)
20	HP33	Yes	/		F	7	hand left	
21	HP21	Yes	/		F	9	distal femur left + right	

		Yes	/				ribs left, humerus right, pelvis right, scapula left, clavicule left, humerus left, hand left	
22	HP2				F	10		
23	HP35	Yes	/		F	10	Femur, tibia ankle foot left	
24	HP17	Yes	Yes	p.G100D (+ / +)	F	18	ilac left, femur left, hand left	hand phalangs digit 5 (CS- I)
25	HP47 HP97	/	Yes		F	18	fibula right	fibula (EC)
26	HP19	Yes	/	p.E546K (na / +)	F	31	hand right, arm right, foot left	
27	HP10	Yes	/		F	36	hand left, wrist left, huemrus left, pelvis, femur left, pelvis	
28	HP5	Yes	/		F	na	hands, feet, arms, legs, pelvis, iliac,	
29	HP58	Yes	/		F		hands	
30	HP18	Yes	/		M	0,9	hand arm foot leg right	
31	HP63	Yes	/		M	1	arms legs left + right	
32	HP16	Yes	/		M	1,08	hand arm right	
33	HP13	Yes	/		M	2	hand arm right	
34	HP72 HP52	Yes	Yes		M	2	hands arms left + right	fibula L (EC)
35	HP40	Yes	/		M	2,5	femur left	
36	HP57	Yes	/		M	3	femur left + right	
37	HP44	Yes	/		M	3	hand right, cubitus right, foot right	
38	HP28	Yes	/		M	3	Feet, hands, legs right + left	
39	HP38	Yes	/		M	4	Hands feet right + left, ankle (maleole)	
40	HP75 HP43	Yes	Yes	p.D30D (+/ +)	M	4	leg left, scapula left	femur R (EC)
41	HP4	Yes	/		M	5	fibulma left, femur left, toes left	
42	HP9	Yes	/		M	5	legs left + right	

43	HP24	Yes	/		M	6,5	fingers, humerus, femur, fibula right	
44	HP36	Yes	/		M	7	ankle left, knees left and right, ribs	
45	HP6	Yes	/		M	8	hip left, ankle left	
46	HP55 HP76	Yes	Yes		M	17	fibula right, femur right, fibula left	fibula R* CS-II; fem R, fib L (EC)

\*Patient HP 17 was the only patient affected with Maffucci syndrome. <sup>1</sup>abbreviations: Dg: diagnosis; f: female; m: male; R: right; L: left; EC: enchondroma; CS: chondrosarcoma; na: non available. Patients HP 11, 17, 19 presented an ovary kyste, patient HP2 had surgery for a tumor of the ovary granulosa at age 13 years; her aunt presented an ovary kyste. That patient (HP2) had also scoliosis. Patient HP33's mother was affected with Scheuermann's disease. Patient B285's father was affected with Fiessinger-Leroy-Reiter syndrome. All other family histories were unremarkable.

Supplementary Table 2: Description of the patients affected with Maffucci syndrome / Ollier disease. Belgium cohort.

	MAF	PTHR1 screening		PTHR1 Variant	Sexe	Birth date day/month/year	Clinical data	
		Blood	Tissue	Tissue/ blood			VM	enchondromas
1	M20	Yes	/	/	M	07 04 /1966	40 on the left hand + 6 on the left forearm + 18 on the left foot. Right leg is approx 6 inches shorter than left leg	1 on the right thumb + 2 on the right femur
2	M50	Yes	Yes	/	F	09 11 /1972	right upper extremity + right lower extremity + trunk	Right : foot, hand, arm, chest/sternum
3	M70	Yes	/	/	M	30 10 /1981	Lip, right hand, chest	hand, trunk (chondrocostal and scapula), proximal femur
4	M90	Yes	Yes	/	M	?	?	?
5	M100	Yes	/	/	M	07 06 /1971	Head/neck, right and left hand, right and left shoulder, foot	right and left upper extremity, left lower extremity (birth and puberty) + Chondrosarcoma on the right shoulder
6	M110	Yes	Yes	/	M	25 03 /1983	VM on the right hand, right arm, right femur	finger and arm, scapula, shinbone, fibula
7	M120	Yes	/	/	F	27 11 /1972	VM on the left hand, left arm, left shoulder, right foot, thigh bone right + spindle cell hemangioma-endothelioma	middle finger, hand, arm, shoulder

8	M140	Yes	Yes	/	M	07 12 /1991	right and left feet, right stomach, right throat, righth buttock	Shoulder, fibula, femur, elbow, foot, wrist, fingers, ribs, tibia
9	M150	Yes	/	/	F	24 12 1993	Right and left hands, right upper eyelid and ear, right and left plantar surface of foot and toes, oral mucosa	on the right leg
10	M160	Yes	Yes	/	M	19 06/1987	VM on right foot	fibula
11	M170	Yes	Yes	/	M	?	?	?
12	M180	Yes	Yes	/	F	22 03/1975	On the sole of the left foot + left leg, pelvic bones, arm, hand, fingers	On the arm, hand and fingers + On the pelvic bones, knee, thigh, shank, foot, leg toes
13	M190	Yes	Yes	/	M	03 05/1956	VM on the right and left trunk	right upper extremity, left upper extrem, trunk, right lower extremity + chondrosarcoma
14	M200	Yes	Yes	/	F	19 04/1988	VM of the ipsilateral skin over scapula	Right hand + forearm
15	M210	/	Yes	/	F	10 07/1989	/	?
16	M230	Yes	/	/	M	23 11/1970	foot, thigh	foot, thigh
17	M240	Yes	/	/	F	25 10/1966	On the hand + internal angiomas (liver)	shoulder, leg, arm and hand + overgrowth of the left leg. Chondr 3° degré
18	M250	Yes	Yes	/	F	?	right foot, right hand, right knee (femur)	Dorsal face of the right hand
19	M260	Yes	Yes	/	M	06 11/1981	Multiple VMs on right ankle and distal tibia, on right foot,	foot, ankle

20	M280	Yes	/	/	F	?	foot	?
21	M290	Yes	Yes	/	F	?	?	?
22	M300	Yes	/	/	F	10 07/1977	VM on the left arm and hand, trunk	bones of left arm, forearm and hand + phleboliths.
23	OLR30	Yes	Yes	A122T 0/+	M	?	/	right lower extremities
24	OLR50	Yes	/	/	F	06 01/1989	/	skull + right lower extrem
25	OLR60	/	Yes	/	M	05 04/1987	/	right and left upper and lower extrem
25	OLR70	Yes	/	/	M	06 11/1991	/	Left lower extrem
27	OLR120	Yes	/	/	F	21 12/1944	/	Right lower extrem
28	OLR130	Yes	/	/	M	28 03/1966	/	Left and right upper extrem
29	OLR140	Yes	/	/	M	25 11/1998	/	Right upper extrem and left lower extrem
30	OLR150	Yes	/	/	M	10 19/1989	/	Left hand
31	OLR160	Yes	/	/	F	27 04/1966	/	Right upper extrem
32	OLR170	Yes	/	/	F	03 12/1997	/	Left upper extrem, trunk, left lower extrem
33	OLR180	Yes	/	/	M	02 01/1991	/	head/neck, left and right upper/lower extrem, trunk
34	OLR190	Yes	Yes	G121E 0/+	F	12 10/1997	/	Left and right upper extrem, left and right extrem and trunk
35	OLR330	Yes	/	/	M	?	/	?
36	OLR340	Yes	/	/	M	17 10/1990	/	Lower extremities
37	OLR350	Yes	/	/	F	?	/	?
38	OLR360	Yes	/	/	F	23 11/2000	/	left leg, right arm, hands, pelvis

MAF: Maffucci, OLR: Ollier, VM: vascular Malformation