

Table S1. A literature review of phacomatosis pigmentokeratolica (PPK) cases published during 1997–2012 using Pubmed

First author	Publication year	Sex/Age, years	Distribution	Extracutaneous anomalies	Other cutaneous lesions
Oh GN (8)	2012	M/0	The face, scalp and right side of the trunk		
Baroni A (9)	2012	M/13	The left side of the body	Scoliosis; Electrocardiography showed right bundle branch block.	Café-au-lait macule
Chantorn R (7)	2011	F/18	The face, tongue, scalp and right side of the trunk		
Taheri AR (10)	2011	M/21	The left side of scalp, face and body		Compound melanocytic naevus of the conjunctiva; longitudinal melanonychia
Tara A (11)	2011	F/25	The left side of the arm and body	Arteriovenous fistulation; left coxarthrosis and impaired longitudinal bone growth in the left leg	
Jacobelli S (12)	2010	M/0.5	The left side of head and body	Juvenile hypertension; nephroblastoma of the left kidney	Basal cell carcinoma; syringo-cystadenoma papilliferum
Polat M (13)	2008	F/39	Left zygoma and left side of the scalp		
Majmudar V (14)	2007	M/44	The right side of body and leg	Headaches and blurred vision; hyperhidrosis; panhypopituitarism; large suprasellar dermoid cyst; left homonymous hemianopia, impaired speech, learning disability, impaired dexterity of the right arm and bilateral pes cavus	Spitz naevus
Gruson LM (15)	2006	M/14	Left side of head to foot	Hemihypertrophy and a rhabdomyosarcoma of the abdominal wall	Large café-au-lait macules; multiple basal cell carcinomas
Bouthors J (5)	2006	M/54	The right side of scalp, face and body	McCune-Albright syndrome;	Spitz naevi; basal cell carcinoma; superficial spreading melanoma
Martinez-Menchón T (16)	2005	M/24	The left side of body and face	Pheochromocytomas; severe rickets	Blue naevus
Okada E (17)	2004	M/18	The left side of body and face	Right-eye limbic tumour of naevocytic origin; retarded cerebral maturation with paroxysmal subcortical dysfunction; bifid L4 arch	
Kinoshita K (18)	2003	M/33	The left side of head and neck	Left eye microphthalmia; juvenile hypertension (renal artery stenosis); kyphoscoliosis; dumb-bell-shaped solid tumours involving the spinal roots between Th11 and L4;	
Hill VA (19)	2003	F/5	Right abdomen, arm and leg down to the foot	arachnoid cyst in the left middle cranial fossa and a lipomatous tumour in the left orbit	Spitz naevi
Wollenberg A (20)	2002	M/23	Both buttocks, the right thigh, the right knee, the right pectoral region and the upper back	Epilepsy; right hemiparesis	
Boente MC (21)	2000	M/9	The left side of scalp, face, arm and body	Hyperhidrosis of the palms, soles and axillae	Café-au-lait macula; melanocytic dysplasia; collagen naevi
		M/9	The left side of head, neck and oral mucosa	Hemiatrophy of the left body side; slight hypothyroidism; hyperhidrosis	
		M/4	The right side of the scalp, neck, extremities and the trunk	Left-sided scoliosis; blind	
Tadimi G (22)	1998	M/32	The right side of the scalp, neck, and the both sides of the trunk	Fragile X chromosome	Melanoma
		F/16	The left side of arm and body	Scoliosis; hemiatrophy and mild strength reduction of the left arm and leg; hyperreflexia of the ankle and knee joints; hyperpathia; hyperhidrosis;	
		M/5	Left head down to the foot	Congenital glaucoma; congenital torticollis; left facial hemiatrophy; scoliosis; intellectual deficit	
Hermes B (23)	1997	F/13	The left side of the body	Conductive hypacusis at the brainstem level; eye abnormalities with esotropia; facial dysmorphism; hypophosphatemic rickets	
		F/10	Body	Hyperpathia; dysesthesia; hyperhidrosis	
				Minor deviation of the spine	

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