

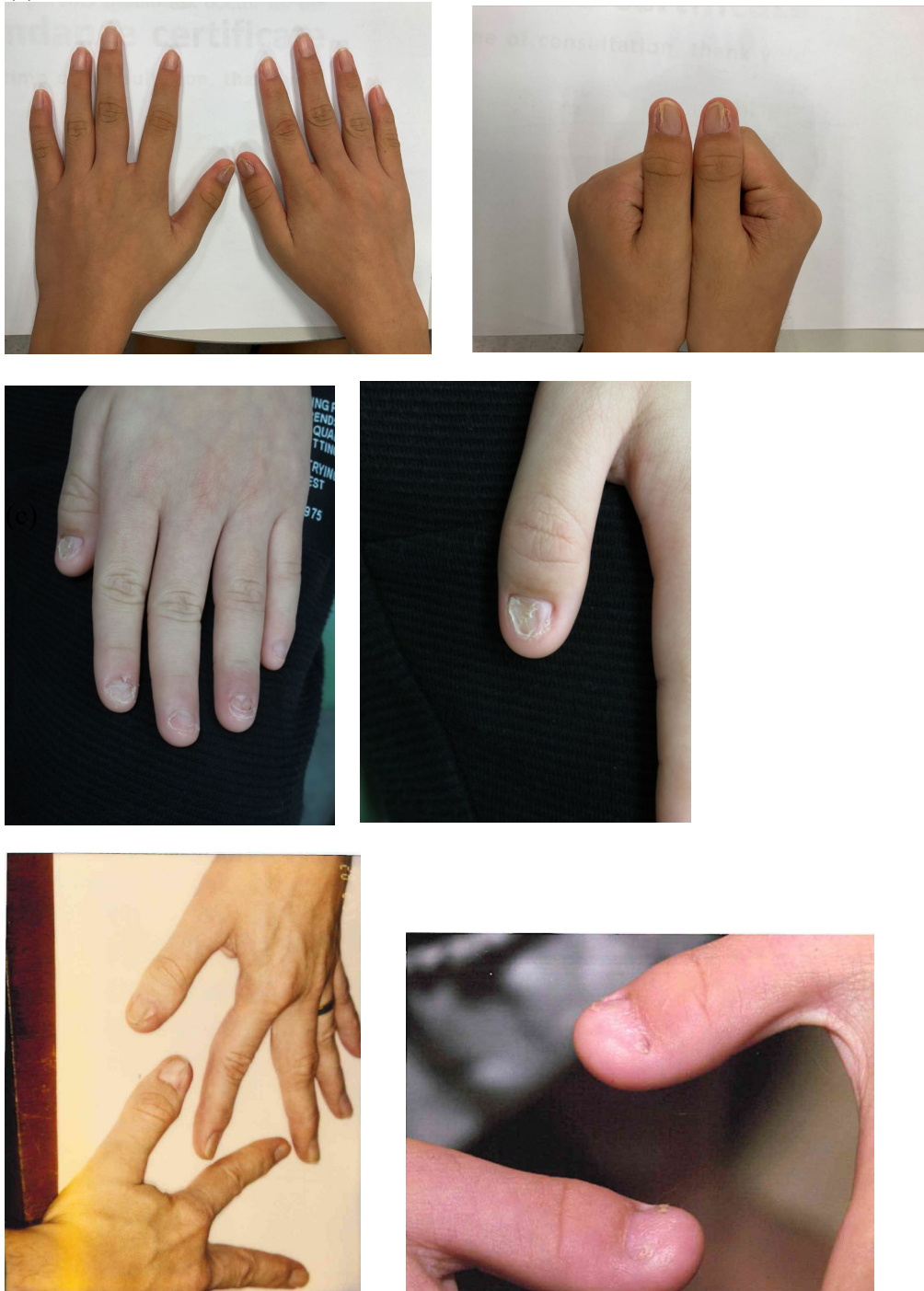
Supplementary material

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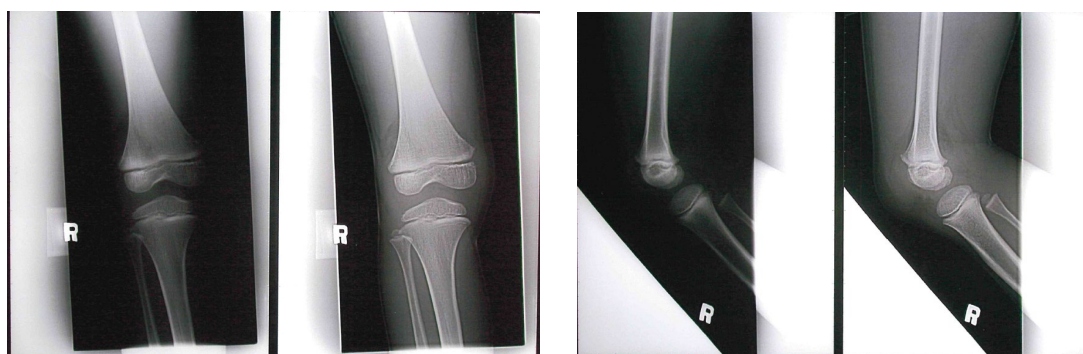
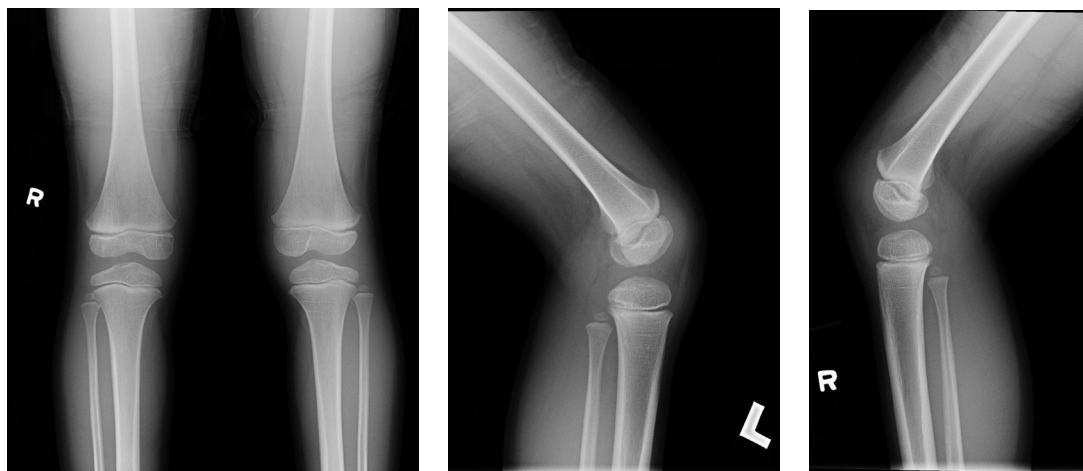
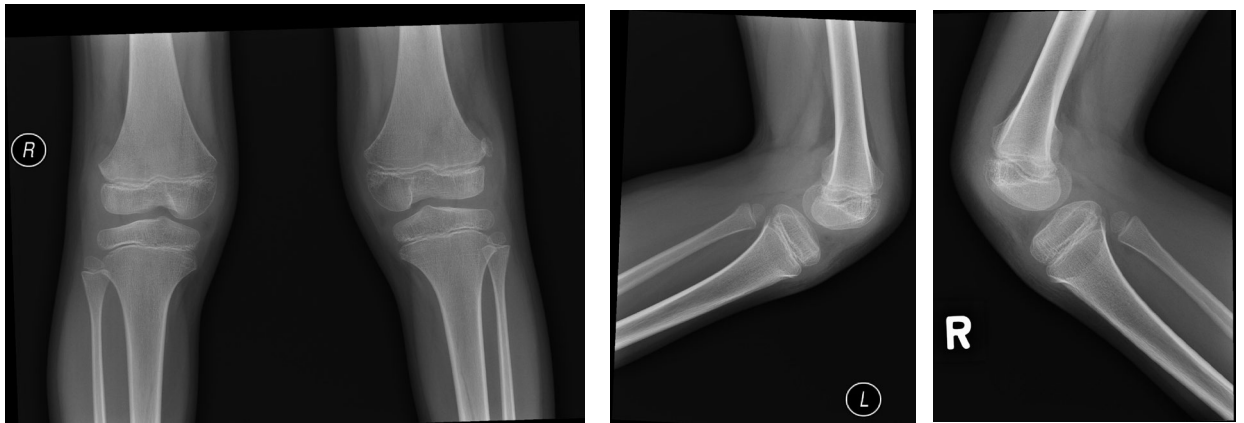
Supplement to: LT Leung, SKL Ho, WC Yiu, et al. Novel *LMX1B* variants in nail-patella syndrome. Hong Kong Med J 2025 Feb;31(1):75-7 | Epub 17 Feb 2025.
<https://doi.org/10.12809/hkmj2411483>.

Supplementary Figure. (a) Dysplastic nails with longitudinal and horizontal splitting and ridges. Among all patients, the thumbnails are most severely affected. (b) X-rays of bilateral knees in anteroposterior, lateral, and skyline views, demonstrating absent or hypoplastic patellae. (c) Hypoplastic patellae with lateral subluxation. (d) A patient with bilateral elbow subluxation and contracture present since birth. (e) X-rays of elbows revealing bilateral hypoplasia, posterior dislocation of radial heads, and bowing deformity of the radius. (f) X-rays of the pelvis with iliac horns

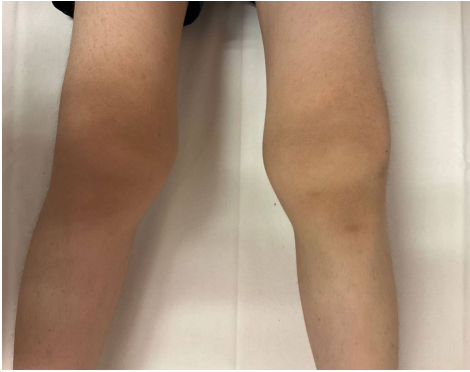
(a)



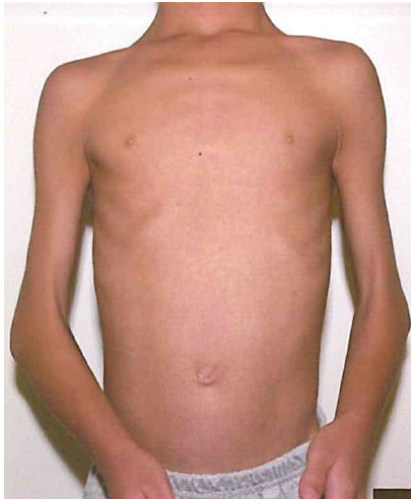
(b)



(c)



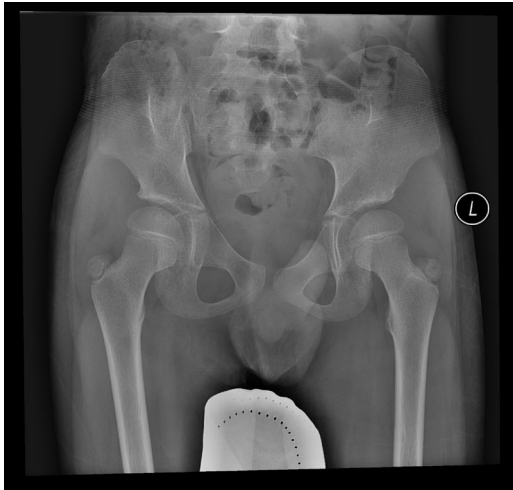
(d)



(e)



(f)



Supplementary Table. Clinical and molecular findings among individuals with nail-patella syndrome

Family	F1	F2		F3	F4	F5	
Individual	1	2	3	4	5	6	7
Sex	M	F	M	M	M	M	M
Age at presentation	7y 3m	1m	N/A	N/A	At birth	N/A	32y 11m
Age at last consultation	13y 3m	11y 11m	45y 1m	54y 3m	26y 2m	74y	47y 3m
Family history	Y	Y	Y	Y	N	Y	Y
Molecular findings							
Variant	c.432C>A	c.745C>T		c.175T>C	Exon 4-6 deletion	Exon 1-2 deletion	Exon 1-2 deletion
Protein change	p.(Cys144Ter)	p.(Arg249Ter)		p.(Cys59Arg)			
Location	Exon 5	Exon 5		Exon 2	Exon 4-6	Exon 1-2	Exon 1-2
Inheritance	N/A	Paternal	N/A	N/A	N/A	N/A	Paternal
Clinical characteristics							
Presenting feature	Frequent falls	Ptosis Corneal opacity	Family screening	Incidental finding on knee X-ray	Bilateral elbow flexion contracture deformity	N/A	Family screening
Nail changes	Triangular lunulae Dystrophic thumbnails with longitudinal splitting	Triangular lunulae Dystrophic thumbnails with horizontal splitting	Dystrophic nails on thumbs and index fingers	Longitudinal ridging of thumb and small toe	Dystrophic nails on thumbs, index fingers, and little toes	Dystrophic nails on thumbs	N/A

Knee involvement	Bilaterally absent patellae	Bilaterally small patellae with subluxation	N/A	Nil	Bilaterally small patellae with subluxation	Bilaterally small patellae	N/A
Elbow involvement	Nil	Nil	Nil	Nil	Bilateral elbow flexion contracture deformity since birth, radial head hypoplasia with posterior dislocation	Limited bilateral elbow movement	N/A
Iliac horns on radiographs	Bilaterally small symmetrical posterior iliac horns	Nil	N/A	N/A	Bilateral iliac horns	Bilateral iliac horns	N/A
Renal involvement	Microscopic haematuria	Nil	N/A	N/A	Nil	N/A	N/A
Ophthalmic involvement	Nil	Bilateral congenital ectropion uvea with glaucoma High myopia Retinal lattice degeneration	Right eye blindness since birth, bilateral glaucoma	N/A	Nil	N/A	Right eye cataract Bilateral high myopia (-7.0 dioptries)

		(right eye)					
		Rhegmatogenous					
		retinal					
		detachment (left					
		eye)					
Body height	10%	50%	N/A	N/A	3%	N/A	N/A
percentiles							
Others					Bowing		
					deformity of		
					the radius		

Abbreviations: F = female; M = male; m = months; N/A = not available; Nil = none; Y = yes; y = years