

Supplementary Table 1: Demographic, clinical, radiological and biochemical characteristics of all prior reported cases of congenital ichthyosis with rickets^{1,4-6,8-23}

Sl. No.	Study (year)	Age (years)/Sex (M/F)	Consanguinity	Collodion membrane at birth	Phenotype of ichthyosis	Skeletal and radiological features	Other features	Baseline Ca ²⁺ , PO ₄ ²⁻ (mg/dL)	Baseline Vitamin D 25(OH)D (ng/mL)	Baseline ALP (IU/L)	Baseline PTH (pg/mL)	Additional points or conclusions
1	Bubna <i>et al.</i> (2014) ⁸	45/M	-	Present	Lamellar ichthyosis	Genu varum Bone scan showing increased tracer uptake in the right femur and left tibia	Seizures since birth Carcinoma hypopharynx	Ca ²⁺ : 6.30	9	291	-	Poorly nourished
2	Ali <i>et al.</i> (2013) ⁹	14/F	Present	Present	Lamellar ichthyosis Three affected siblings without rickets	Bowing of upper limbs, genu varum, pectus carinatum Radiography showing greenstick fracture in meta-diaphyseal end of fibula DEXA scan: marked reduced bone mineral density Radionucleotide skeletal scintigraphy: increased uptake and expansion of epiphyseal plates		Ca ²⁺ : 6.60	7.80	1192	354	-
3	Kaushik <i>et al.</i> (2023) ¹⁰	In 56 patients mean age 10.87 ± 8.90; 31/56: M 25/56: F	-	-	Non-syndromic congenital ichthyosis	-	Vitamin D receptor gene expression decreased in skin of ichthyosis patients	Ca ²⁺ : 9.65 ± 0.67 PO ₄ ²⁻ : 5.93 ± 0.75	15.71 ± 11.72	56.12 ± 27.98	48.61 ± 25.24	Values given in ng/mL instead of pg/mL, making converted value of vitamin D at 48,610 pg/mL
4	Sethuraman <i>et al.</i> (2015) ¹¹	In 119 patients, mean age 7.7 ± 4.6; 74/119: M 45/119: F	-	-	In 56/119: autosomal recessive congenital ichthyosis (ARCI) In 24/119: epidermolytic ichthyosis (EI)	In 27 patients, genu varum, pectus carinatum, swelling over wrists and knees In 74 patients that consented for radiological screening, 31 showed evidence of rickets	-	<u>ARCI</u> Ca ²⁺ : 8.9 ± 1.61 PO ₄ ²⁻ : 5.14 ± 2.52 <u>EI</u> Ca ²⁺ : 9.55 ± 2.08	<u>ARCI</u> 8.7 ± 5.75 <u>EI</u> 10.00 ± 7.61 <u>CI</u> 9.37 ± 5.94	<u>ARCI</u> 656.5 ± 514.1 0 682.8 ± 717.6 7 <u>CI</u>	<u>ARCI</u> 95.45 ± 86.77 <u>EI</u> 123.1 ± 155.0 3 <u>CI</u>	-

					In 35/119: common ichthyosis (CI) In 4/119: Netherton syndrome and trichothiodystrophy	With clinical and radiological assessments, a combined 47 patients showed evidence of rickets		PO4 ²⁻ : 5.16 ± 2.63 CI Ca ²⁺ : 9.09 ± 1.33 PO4 ²⁻ : 4.98 ± 1.53		420.3 ± 370.38	62.90 ± 64.12	
5	Chouhan <i>et al.</i> (2012) ¹	In 45 patients with disorders of keratinization (including psoriasis) mean age 8.22 ± 5.2 years; 29/45: M 16/45: F	-	-	EI, non-bullous form erythroderma (NBIE), lamellar ichthyosis, Ichthyosis vulgaris, X-linked recessive ichthyosis	In nine patients, bowing of upper and lower limbs, genu valgum, pectus carinatum, other thoracic and spinal deformities, skeletal dysplasia In 19 patients, radiological changes of rickets seen	-	Ca ²⁺ : 9.30 ± 1.30 PO4 ²⁻ : 4.90 ± 1.40	8.40 ± 5.20	816.80 ± 689.60	110.90 ± 128.20	-
6	Thacher <i>et al.</i> (2004) ¹²	1/M	-	-	Lamellar ichthyosis One affected sibling	Rachitic rosary Radiography showing marked fraying and cupping of distal metaphyses at wrists and knees		Ca ²⁺ : 8.90 PO4 ²⁻ : 2	<4.50	707	63	Prior calcium replacement received for 6 months
		8/M	Present	-	Lamellar ichthyosis	Rachitic rosary, swelling of wrist and ankle Radiography showing longitudinal widening of epiphyseal plates and fraying of distal metaphyses at wrist and knees	-	Ca ²⁺ : 6.90 PO4 ²⁻ : 3.90	<4.50	3941	39	
		7/F	-	-	Epidermolytic hyperkeratosis (EI)	Genu varum Radiographs of wrists and ankles confirm rickets		Ca ²⁺ : 6.80 PO4 ²⁻ : 3	-	987	173	Clinically EI, histopathology is suggestive of X-linked ichthyosis
7	Sethuraman <i>et al.</i> (2016) ⁵	3/M	-	-	ARCI	-	-	Ca ²⁺ : 10.30 PO4 ²⁻ : 3.72	<4	393.50	239	-

		8/F	-	-	ARCI	-	-	Ca ²⁺ : 10 PO ₄ ²⁻ : 6.10	<4	494	635	-
		2.5/M	-	-	ARCI	-	-	Ca ²⁺ : 8.97 PO ₄ ²⁻ : 2.43	<4	285.3 0	152	-
		3/M	-	-	EI	-	-	Ca ²⁺ : 6.70 PO ₄ ²⁻ : 3.6	<4	2998	398	-
		1.5/M	-	-	ARCI	-	-	Ca ²⁺ : 9.12 PO ₄ ²⁻ : 2.60	<4	649.8 0	584	-
		3.5/F	-	-	EI	-	-	Ca ²⁺ : 9.10 PO ₄ ²⁻ : 5.10	<4	268	55.60	-
		0.75/M	-	-	ARCI	-	-	Ca ²⁺ : 10.50 PO ₄ ²⁻ : 4.60	-	446	144	-
8	Angmo <i>et al.</i> (2016) ¹³	16/M	Present	-	Lamellar ichthyosis	Genu valgum Radiography showing widening, cupping and fraying of distal femoral, radial and ulnar metaphyses, osteopenia	Juvenile open angle glaucoma and axial myopia	Ca ²⁺ : 8.20	-	4697	-	-
9	Kothari <i>et al.</i> (2013) ¹⁴	8/M	Absent	-	EI	Genu varum, bowing of right upper limb, deformity of left upper limb (due to an old fracture), scoliosis Radiography showing marked cupping and fraying of distal metaphysis, osteopenia, pseudo- fracture and malunion	-	Ca ²⁺ : 7.20	11.30	1396. 80	-	-
		14/F	Absent	-	Lamellar ichthyosis One affected sibling	Genu valgum, rachitic rosary and swelling of wrists and ankles Radiography showing marked cupping and fraying of distal metaphysic, osteopenia, pseudo-	-	Ca ²⁺ : 7.79	12.25	3090	530	-

						fracture and malunion						
10	Zaouak <i>et al.</i> (2019) ¹⁵	13/M	Present	Present	Lamellar ichthyosis	Genu varum Radiography showing osteopenia, cupping, splaying and fraying of distal metaphysis of upper and lower limbs with loss of provisional zone of calcification and wrists and knees	-	Ca ²⁺ : 23.42	3.61	650	elevated	-
11	Tewari <i>et al.</i> (2020) ¹⁶	5/F	-	Present	Lamellar ichthyosis	Radiography showing osteopenia, distal femoral and radial metaphyseal splaying with irregular and widened physes	Periodontal breakdown and premature loss of primary teeth	Ca ²⁺ : 15.36	1.92	-	-	Only radiological changes mentioned
12	Holm AL <i>et al.</i> (1991) ¹⁷	30/F	-	-	ARCI	Genu varum Radiography showing osteoporosis, multiple healed fractures	impetigo herpetiformis during early third trimester of pregnancy	Ca ²⁺ : 30.99 PO ₄ ²⁻ : 9.72	-	197	-	-
13	el-Khateeb (2008) ¹⁸	8/M	Absent	-	EI	Pectus carinatum, rachitic rosary Radiography showing epiphyseal thickening and other rachitic changes	-	Ca ²⁺ : 8.40	0.10	1435	-	-
14	Taskin <i>et al.</i> (2007) ¹⁹	4/M	Absent	-	Chanarin-Dorfman syndrome Affected sibling and cousin	Rachitic rosary Radiology showing cupping and fraying of distal metaphyseal radius and ulna	Failure to thrive, hepatomegaly, cognitive impairment and delayed motor function, lipid vacuoles in leukocytes	Ca ²⁺ : 5.50 PO ₄ ²⁻ : 5.50	-	1181	281	-
15	Kumar <i>et al.</i> (2007) ²⁰	5/M	Absent	-	Lamellar ichthyosis	Frontal bossing, genu varum, bowing of upper limbs, swelling over wrist, rachitic rosary Radiography showing bowing and shortening with multiple fractures of upper and lower limbs	-	-	5	-	-	-

16	Deka <i>et al.</i> (2012) ⁶	22/F	Present	Present	Lamellar ichthyosis	Genu valgum Radiography showing osteopenia DEXA scan osteopenia	-	Ca ²⁺ : 8.80 PO ₄ ²⁻ : 3.20	9.12	485	68.80	-
17	Nayak <i>et al.</i> (2006) ²¹	6/M	Present	-	EI	Rachitic rosary, swelling of wrists and ankles Radiography showing marked splaying and cupping of the distal metaphyses and widening of the growth plate of the upper end of tibia and fibula and bowing	-	Low	Low	High	High	-
18	Sethuraman <i>et al.</i> (2008) ⁴	12/M	Absent	-	Lamellar ichthyosis One affected sibling	Genu varum, pectus carinatum, rachitic rosary Radiology showing osteopenia, widening, cupping and fraying of the metaphysis, deformities, of femur and tibia, widening of pelvic bones	-	Ca ²⁺ : 5.70 PO ₄ ²⁻ : 4.10	32	2262	53	ALP levels are given in IU/mL
		12/M	-	-	Lamellar ichthyosis	-	-	Ca ²⁺ : 6.70 PO ₄ ²⁻ : 5.10	<5	1416	213	
		7/M	-	-	NBIE	-	Psoriasis with atopy	Ca ²⁺ : 8.30 PO ₄ : 3.30	<5	1443	206.80	
		14/M	-	-	Lamellar ichthyosis	-	-	Ca ²⁺ : 8 PO ₄ ²⁻ : 3.10	-	2215	High	
		9/M	-	-	Lamellar ichthyosis	-	-	Ca ²⁺ : 8 PO ₄ ²⁻ : 5.70	-	1253	-	
19	Barraud <i>et al.</i> (2015) ²²	9.5/-	Absent	-	Chanarin-Dorfman syndrome	Genu valgum Radiography showing metaphyseal anomalies, enlargement of growth plates, fringed appearance, osteomalacia and cup-shaped enlargement of costochondral junction	Arthrogryposis of all limbs with feet in equines varus, axial hypotonia, hepatomegaly, lipid vacuoles in muscle fibres,	Ca ²⁺ : 38	2.88	716 IU	25.52	Article translated from French values may differ

							cognitive impairment and delayed motor function					
20	Neema <i>et al.</i> (2015) ²³	6/F	Absent	-	NBIE	After acitretin, Genu valgum; Radiography: cupping and fraying of distal metaphyses of upper and lower limbs	Acitretin started at 25 mg/day for six months, following which rickets developed Pseudoainhum of fingers	After acitretin for six months Ca ²⁺ : 9.20	After acitretin for six months 6.34	After acitretin for six months 960	-	Biochemistry before starting acitretin: normal
		2/F	Absent	Absent	Lamellar ichthyosis	-	Isotretinoin started at 10 mg/day for six months	-	Before isotretinoin 51.91 After isotretinoin 9.22	-	-	-
21	Our case (2024)	4/M	Absent	Present	NBIE	Radiography of lower limbs showing features of severe bone softening Medullary widening and thinning of cortices of ends of long bones Metaphyseal cupping	Ectropion Photosensitivity	Ca ²⁺ : 8.18 PO ₄ ²⁻ : 3.8	<3	1131	173.90	

Ca²⁺, Calcium; PO₄²⁻ Phosphate; 25(OH)D, 25-hydroxycholecalciferol; ALP, Alkaline Phosphatase; IU, International Units; PTH, Parathyroid hormone; DEXA, Dual energy X-ray absorptiometry; ARCI, Autosomal recessive congenital ichthyosis; EI, Epidermolytic ichthyosis; CI, Common ichthyosis.

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