

**Table SII. Patients affected by autosomal recessive congenital ichthyosis: demographics, clinical features, and family occupational status**

Variable	n (%)
Overall	94 (100)
Sex	
Male	41 (43.6)
Female	53 (56.4)
Age,	
< 18 years	52 (55.3)
≥ 18 years	42 (44.7)
Clinical type	
Lamellar <sup>a</sup>	80 (85.1)
Erythrodermic	14 (14.9)
Mutated gene	
Undetermined	45 (46.8)
<i>ABCA12</i>	6 (6.4)
<i>ALOX12B</i>	11 (11.7)
<i>ALOXE3</i>	4 (4.3)
<i>CYP4F22</i>	5 (5.3)
<i>NIPAL4</i>	4 (4.3)
<i>SDR9C7</i>	1 (1.1)
<i>TGM1</i>	18 (20.2)
Sign/symptom score	
0–5	71 (75.5)
≥ 6	23 (24.5)
Ear deformity	
No	76 (80.9)
Yes	18 (19.1)
Large scales	
No	53 (56.4)
Yes	41 (43.6)
Fissures	
No	66 (70.2)
Yes	28 (29.8)
Itch	
No	14 (14.0)
Yes	80 (85.1)
Recurrent infections	
No	76 (80.9)
Yes	18 (19.1)
Foul-smell	
No	63 (67.0)
Yes	31 (33.0)
Walking problems	
No	81 (86.2)
Yes	13 (13.8)
Ectropion	
No	67 (71.3)
Yes	27 (28.7)
Palmoplantar keratoderma	
No	30 (31.9)
Yes	64 (68.1)
Visits/year	
1	48 (51.1)
2–3	28 (29.8)
≥ 4	18 (19.1)
Mother works <sup>b</sup>	
No	43 (55.1)
Yes	35 (44.9)
Father works <sup>b</sup>	
No	9 (12.0)
Yes	66 (88.0)
Workdays lost by caregivers <sup>b</sup>	
No	24 (27.9)
Yes	62 (72.1)

<sup>a</sup>Includes 2 patients with harlequin ichthyosis. <sup>b</sup>Totals may vary due to missing values. Lamellar: lamellar ichthyosis; Erythrodermic: congenital ichthyosiform erythroderma.