

Unknown mutations and genotype/phenotype correlations of autosomal recessive congenital ichthyosis in patients from Saudi Arabia and Pakistan

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Table S1. Homozygosity regions matching ARCI gene intervals identified after homozygosity mapping.

Patient	Chromosome region	% Heterozygosity	Length	Cytoband	ARCI gene match
SA-01	chr14:23,172,456-25,025,737	0.0	1,853,282	14q11.2 - q12	<i>TGM1</i>
SA-02	chr2:202,252,090-215,853,470	0.304	13,601,381	2q33.1 - q35	<i>ABCA12</i>
SA-04	chr14:22,879,230-28,961,059	0.630	6,081,830	14q11.2 - q12	<i>TGM1</i>
SA-05	chr14:23,172,456-25,203,263	0.0	2,030,808	14q11.2 - q12	<i>TGM1</i>
SA-06	chr2:175,291,934-220,342,826	0.172	45,050,893	2q31.1 - q35	<i>ABCA12</i>
	chr14:19,283,777-29,454,152	0.283	10,170,376	14q11.2 - q12	<i>TGM1</i>
SA-08	chr5:148,741,029-168,469,088	0.0	19,728,060	5q33.1 - q35.1	<i>NIPAL4</i>
SA-09	chr5:140,154,033-161,846,378	0.118	21,692,346	5q31.3 - q34	<i>NIPAL4</i>
	chr15:91,645,431-100,338,915	0.0	8,693,485	15q26.1 - q26.3	<i>CERS3</i>
SA-10	chr5:141,611,117-169,435,977	0.0	27,824,861	5q31.3 - q35.1	<i>NIPAL4</i>
SA-11	chr5:123,557,892-159,370,345	0.072	35,812,454	5q23.2 - q33.3	<i>NIPAL4</i>
SA-12	chr14:21,340,759-32,111,277	0.105	10,770,519	14q11.2 - q13.1	<i>TGM1</i>
SA-13	chr14:19,283,777-32,590,349	0.226	13,306,573	14q11.2 - q13.1	<i>TGM1</i>
SA-14	chr6:25,601,239-50,108,951	0.037	24,507,713	6p22.2 - p12.3	<i>PNPLA1</i>
	chr17:6,908,440-14,157,308	0.089	7,248,869	17p13.1 - p12	<i>ALOXE3/ALOX12B</i>
	chr14:20,866,317-25,502,331	0.0	4,636,015	14q11.2 - q12	<i>TGM1</i>
SA-15	chr19:15,434,569-22,399,457	0.206	6,964,889	19p13.12 - p12	<i>CYP4F22</i>
YE-01	chr14:24,679,877-32,924,012	0.77	8,244,135	14q11.2 - q12	<i>TGM1</i>
PK01-01	chr2:179,263,031-220,406,107	aggregate of 3 patients NA	41,143,076	2q31.2 - q35	<i>ABCA12</i>
PK01-02					
PK01-04					
PK02-01	chr2:186,002,691-223,060,424	aggregate of 2 patients NA	37,057,733	2q32.1 - q36.1	<i>ABCA12</i>
PK02-02	chr5:149,594,981-163,460,290				
PK03-01	chr17:770,461-13,845,956	aggregate of 2 patients NA	13,075,495	17p13.3 - p12	<i>ALOXE3/ALOX12B</i>
PK03-04					
PK04-01	chr17:5,757,535-13,141,886	0.094	7,384,352	17p13.2 - p12	<i>ALOXE3/ALOX12B</i>
PK05-01	chr5:149,650,749-166,821,863	aggregate of 3 patients NA	17,171,114	5q33.1 - q34	<i>NIPAL4</i>
PK05-03					
PK05-04					