#80 - PREVALENCE OF GENOTYPES RELATED TO LACTOSE INTOLERANCE AND VITAMIN D DEFICIENCY IN CHILEAN PATIENTS WITH IBD

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Background. Lactose intolerance (LI) affects 33% to 75% of the world's population. LI can be secondary to primary or secondary lactose malabsorption. The single-nucleotide polymorphism (SNP) of the lactase gene (LCT) rs4988235-CC confers the non-persistence of lactase. Studies have reported a prevalence of 57% of the CC genotype in the Hispanic Chilean population and 88% in Amerindians, with an important correlation with symptoms of intolerance to the digestion of dairy products. This group of patients might avoid dairy consumption. Patients with IBD may have insufficient production of lactase. In addition, vitamin D deficiency has been reported in patients with IBD. Aim. To evaluate the prevalence of rs4988235-C in Chilean patients with IBD and genotypes associated with vitamin D deficiency (VDD). Methods. 192 patients with IBD were genotyped using Illumina screening Array, the prevalence of the VDD genotypes rs4988235-CC, rs12785878-GG, rs2282679-CC, and rs10741657-GG were evaluated. Results. Table 1 shows the prevalence's of the genetic variant related to Lactose Intolerance and Vitamin D Deficiency in IBD Chilean Patients. The frequency of the C allele for rs4988235 for Chilean IBD patients was 0.79, similar to general American Population (Mexican, Puerto Rican, Peruvian, and Colombian) and Chilean =0.78, and 0.77, and higher than European =0.49).

Conclusion: More than half of Chilean IBD patients have a genetic predisposition to lactose malabsorption, and 5-44% have genetic variants linked to low vitamin D levels. Screening for lactose intolerance and vitamin D deficiency is crucial in this population to prevent negative health outcomes, including an increased risk of osteoporosis. Lactose intolerance should be considered in symptomatic Chilean IBD patients with inactive disease. The lactase genotype rs4988235-CC is prevalent in the American population, suggesting that lactose intolerance may be a issue for Latin American IBD patients.

Table 1.Frequency Genetic Variants related to Lactose Intolerance and Vitamin D Levels in Chilean IBD Patients

Risk Variant	Chr	Gene	Risk Allele		Allele Frequency	Phenotype	Genotype		
					C=0.79		TT	TC	CC
rs4988235	2	LCT	C	Encodes for the lactase enzyme	T=0.21	Lactose Malabsortion	5(2.6%)	70(36.4%)	117(61%)
				Encodes human group-specific component, which is	C=0.22		CC	CA	AA
rs228679	4	GC	C	the major vitamin D-binding protein (VDBP) in plasma	A=0.88	Deficiency Vit D	10(5.2%)	65(33.8%)	117(61%)
				Gene that produces the 7-dehydrocholesterol	G=0.42		GG	GT	TT
				reductase enzyme, which catalyses the production					
rs12785878	11	DHCR7	G	of cholesterol from 7-Dehydrocholesterol using NADPH	T=0.58	Deficiency Vit D	37(19.3%)	88(45.8%)	67(34.9%)
					G=0.67		AA	AG	GG
				CYP2R1 gene has been linked by several studies to					
rs10741657	11	CYP2R1	G	vitamin D serum concentrations	A=0.33	Deficiency Vit D	20(10.4%)	86(44.8%)	86(44.8%)