The prevalence of lactose intolerance (adult hypolactasia) in a randomly selected New Zealand population

Lactose intolerance (adult-type or primary hypolactasia) in Caucasians is predominantly determined by a single nucleotide polymorphism (SNP) in intron 13 of the MCM6 helicase gene, 13,910 bases upstream of the first base of the lactase gene on chromosome 2. The presence of a cytosine (C) at this site confers intolerance to lactose. This is a recessive trait, so the SNP combinations C/T and T/T are lactose-tolerant, while C/C confers the intolerant phenotype. The C to T transition is of relatively recent origin (10,000 to 12,000 years ago), and was strongly selected for with the advent of herding and the associated activity of consuming bovine milk.

This mutation arose in a northern European population, and is thus widely distributed in their descendants. Nearly all of the Caucasian New Zealand population derives from northern Europe and would be expected to be in large part lactose tolerant, but the prevalence of primary (inherited) lactose intolerance in the New Zealand adult population has yet to be formally documented.

Method—Rapid lysis DNA samples were genotyped in batches of 50. A PCR mastermix containing buffer, dNTPs, flanking primers and Platinum Taq DNA polymerase (Invitrogen) was prepared and dispensed in 9 µL aliquots. 1.0 µL of sample DNA was added to each 9 µL reaction, and the samples amplified in an Eppendorf MasterCycler using standard cycling parameters. Following amplification, a cocktail containing restriction endonuclease BsmF1 (New England Biolabs, 2000U/mL) was prepared, and 15 µL added to each sample. Digestion was carried out at 65°C for 120 min, followed by heat inactivation at 80°C for 30 min. Digests (8 µL) were electrophoresed on 15×15 cm² 3% agarose gels, stained in ethidium bromide solution, and visualised under UV light.

Results and Discussion—The numbers of the three genotypic combinations in this population (n=1064) was 88 C/C homozygotes (lactose intolerant), 419 C/T heterozygotes, and 557 T/T homozygotes. Thus, the percentage of this population that is genetically lactose intolerant is 8.23%. The allele frequencies found are consistent with the expected Mendelian distribution (p=0.72 and q = 0.28).

The finding that more than 8% of this sample of the Christchurch population has adult type hypolactasia may be surprising, but it is consistent with data on lactose intolerance in other countries. General practitioners should be wary when patients complain of non-specific symptoms associated with the ingestion of dairy products. The existence of this simple and rapid genetic test to determine the presence or absence of adult hypolactasia should make the task of gastroenterologists to distinguish between primary and secondary lactose intolerance considerably easier.

A substantial majority of southeast Asian peoples are lactose intolerant (among Thais, the figure is nearly 100%), and the influx of Asians into the Christchurch urban region in recent years will have increased the incidence of adult type hypolactasia. Of the thirty survey participants self-identifying as being either Māori or Polynesian (2.9%
of the sampled population), 9 were found to be lactose intolerant. This frequency of 30% is highly statistically significant ($p=0.0004$, Fisher’s exact test).

The finding of a significantly increased prevalence of lactose intolerance in the Polynesian and Māori communities is consistent with current theories about the origin of these people (in Southeast Asia), but requires more formal documentation. It is hoped that this can be achieved in the near future.

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Acknowledgements: The authors thank Drs Michael Burt, Bruce Chapman, and Judith Collett of the Department of Gastroenterology, Canterbury District Health Board for granting access to the bank of DNA samples.

References: