Human gastrin (GAS) RFLP recognised by digestion with BamHI or EcoRI

M.A. Schmidt, G.E. Woloschak, V.V. Michels, J.A. Lust and H. Gordon

Department of Medical Genetics, Division of Immunology and Division of Hematology and Internal Medicine, Mayo Clinic and Mayo Foundation, Rochester, MN 55905, USA

SOURCE AND DESCRIPTION OF CLONE: The human gastrin gene cDNA insert in clone pHG529 (1) is 463 bp in length. This excludes the poly (dC) tails and includes 8 nucleotides of 5' untranslated region, the entire coding region of 303 nucleotides, a 3' untranslated region of 102 nucleotides and a poly (A) track of 50 residues.

POLYMORPHISM: Digestion with BamHI revealed a two allele polymorphism with allele 1 defined by a single band 2.0 kb in size and allele 2 by a single band 2.4 kb in size. EcoRI digestion showed a similar two allele polymorphism. Allele 1 is defined by a band 2.6 kb in size while allele 2 is a band 3.0 kb in size.

FREQUENCY FOR POPULATION STUDIED: Studied in 17 Caucasians with neurofibromatosis, 7 of their unaffected family members and 3 normal controls. The following frequencies were calculated on the basis of the 17 affected and 3 controls. BamHI: allele 1 was 0.95 whereas allele 2 was 0.05. Similarly, EcoRI allele 1 was 0.95 and allele 2 was 0.05.

OTHER ENZYMES TESTED: Not polymorphic for HaeIII in all of the above.

CHROMOSOMAL LOCATION: Human Chromosome 17cen–qter (2).

MENDELIAN INHERITANCE: Co-dominant segregation has been shown for BamHI and EcoRI polymorphisms in one family, Figure 1.

PROBE AVAILABILITY: The pHG529 probe was kindly provided by Dr. Ove Wiborg, University of Aarhus, Denmark.

OTHER COMMENTS: Insert was excised and oligolabeled with 32P-dCTP at room temperature overnight. Final wash was in 0.1X SSC / 0.2% SDS at 60°C for 20 minutes.


Present addresses: Division of Biological and Medical Research, Argonne National Laboratory, Argonne, IL, USA

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Figure 1