

Diabetes tipo II

- Background genético
- Influência ambiental e comportamental.

Table 1 Novel T2D-associated loci						
Novel Locus	Chr	rsID--Risk Allele	OR (95% CI) P-value			MAF
			Stage1 Discovery Meta-analysis	Stage2 Replication Meta-analysis	Stage1 + Stage2 Combined Meta-analysis	
LYPLAL1/ZC3H11B (1q41)	1	rs2820443-T	1.08 (1.04-1.13) 2.94 × 10 ⁻⁴ a	1.06 (1.03-1.09) 2.10 × 10 ⁻⁵ b	1.07 (1.04-1.09) 2.56 × 10 ⁻⁸ c	0.28
EHMT2 (6p21.33-p21.32)	6	rs115884658-A	1.34 (1.18-1.53) 1.00 × 10 ⁻⁵ a	1.17 (1.09-1.26) 2.90 × 10 ⁻⁶ c, d	1.21 (1.14-1.29) 3.00 × 10 ⁻¹⁰ c	0.02
ABO (9q34.2)	9	rs505922-C	1.07 (1.03-1.11) 6.93 × 10 ⁻⁴ a	1.06 (1.03-1.09) 1.90 × 10 ⁻⁵ b	1.06 (1.04-1.09) 4.94 × 10 ⁻⁸ c	0.34
NEUROG3 (10q22.1)	10	rs2642587-G	1.12 (1.08-1.16) 8.45 × 10 ⁻⁹ e	-	-	0.22
CAMKK2 (12q24.31)	12	rs3794205-G	1.09 (1.05-1.14) 4.18 × 10 ⁻⁵ a	1.06 (1.03-1.09) 1.60 × 10 ⁻⁴ b	1.07 (1.04-1.10) 4.11 × 10 ⁻⁸ c	0.32
CALCOCO2/ATP5G1/UBE2Z/SNF8/GIP (17q21.32)	17	rs12453394-A	1.08 (1.04-1.12) 7.86 × 10 ⁻⁵ a	1.07 (1.03-1.11) 9.60 × 10 ⁻⁵ b	1.07 (1.05-1.10) 3.23 × 10 ⁻⁸ c	0.47
AGTR2 (Xq23)	X	rs146662075-T	3.09 (2.06-4.60) 3.24 × 10 ⁻⁸ f	1.57 (1.19-2.07) 1.42 × 10 ⁻³ g	1.95 (1.56-2.45) 7.85 × 10 ⁻⁹	0.008

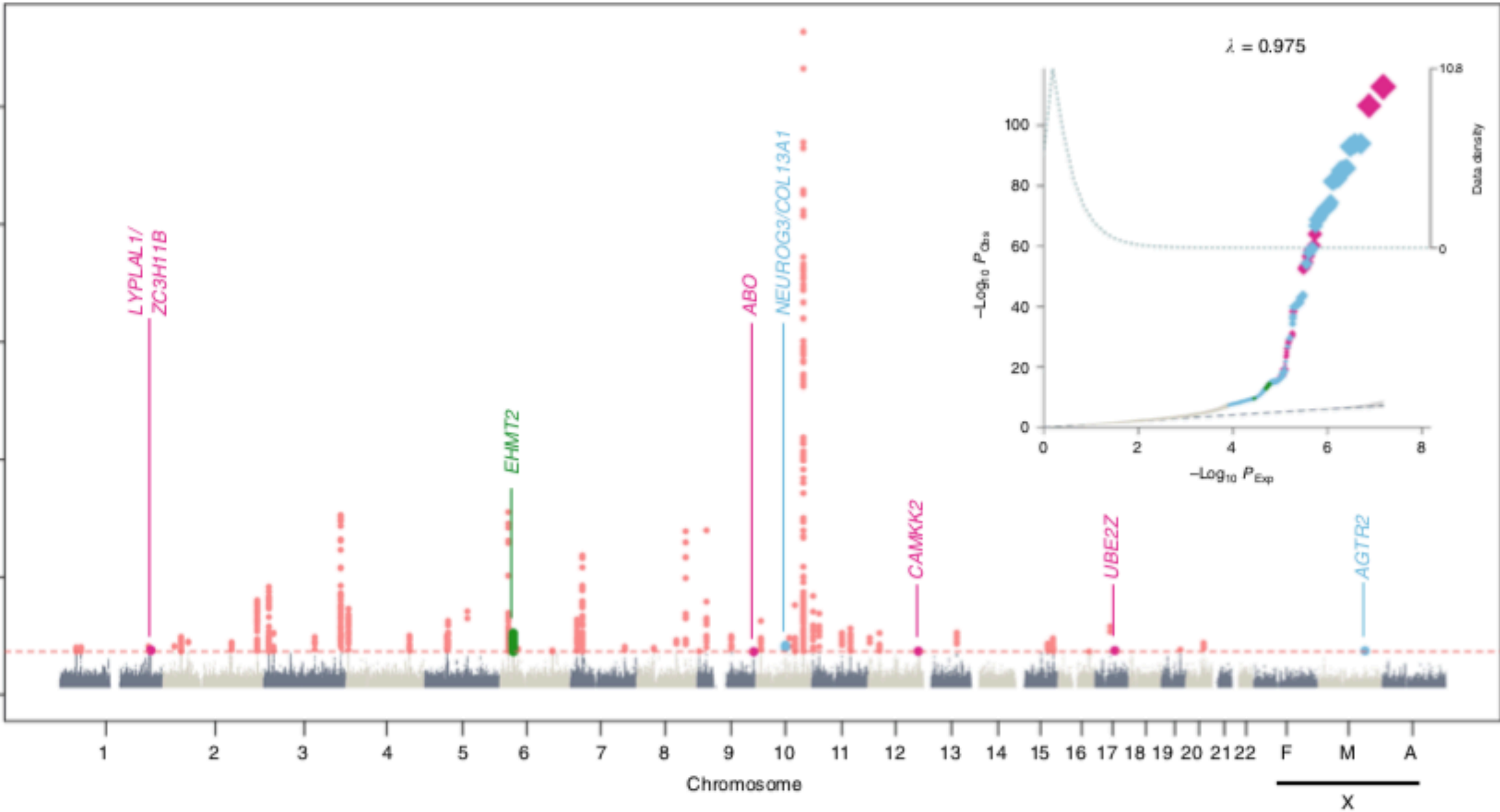
Chr chromosome, OR odds ratio, MAF minor allele frequency

^aImputed based public GWAS discovery meta-analysis (NuGENE + GERA cohort, 7,522 cases and 50,446 controls)

^bTransancestry DIAGRAM Consortium (26,488 cases and 83,964 controls)^cMeta P-value estimated using a weighted Z-score method due to unavailable SE information from Stage 2 replication cohorts^dT2D Diabetes Genetic Portal (Exome-Chip + Exome Sequencing, 35,789 cases and 56,738 controls)^eFull imputed based public GWAS meta-analysis (NuGENE + GERA cohort + GENEVA + FUSION + WTCCC, 12,931 cases and 57,196 controls)

^f70KforT2D Men Cohort (GERA cohort + GENEVA + FUSION, 5,277 cases and 15,702 controls older than 55 years)

^gReplication Men Cohort SIGMA UK10K imputation + InterAct + Danish Cohort (case control and follow-up) + Partners Biobank + UK Biobank (18,370 cases and 88,283 controls older than 55 years and OGTT > 7.8 mmol l⁻¹, when available)



- Sequenciaram 1092 indivíduos de diversas populações.
- Identificação das variações genéticas encontradas.
- Frequências por população.
- Impacto.
- 2012

Table 1: GWAS for common diseases and traits

Phenotype	Number of GWAS loci	Proportion of heritability explained (%)*
Type 1 diabetes	41	~60
Fetal haemoglobin levels	3	~50
Macular degeneration	3	~50
Type 2 diabetes	39	20–25
Crohn's disease	71	20–25
LDL and HDL levels	95	20–25
Height	180	~12

ARTICLE

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An integrated map of genetic variation from 1,092 human genomes

The 1000 Genomes Project Consortium*

