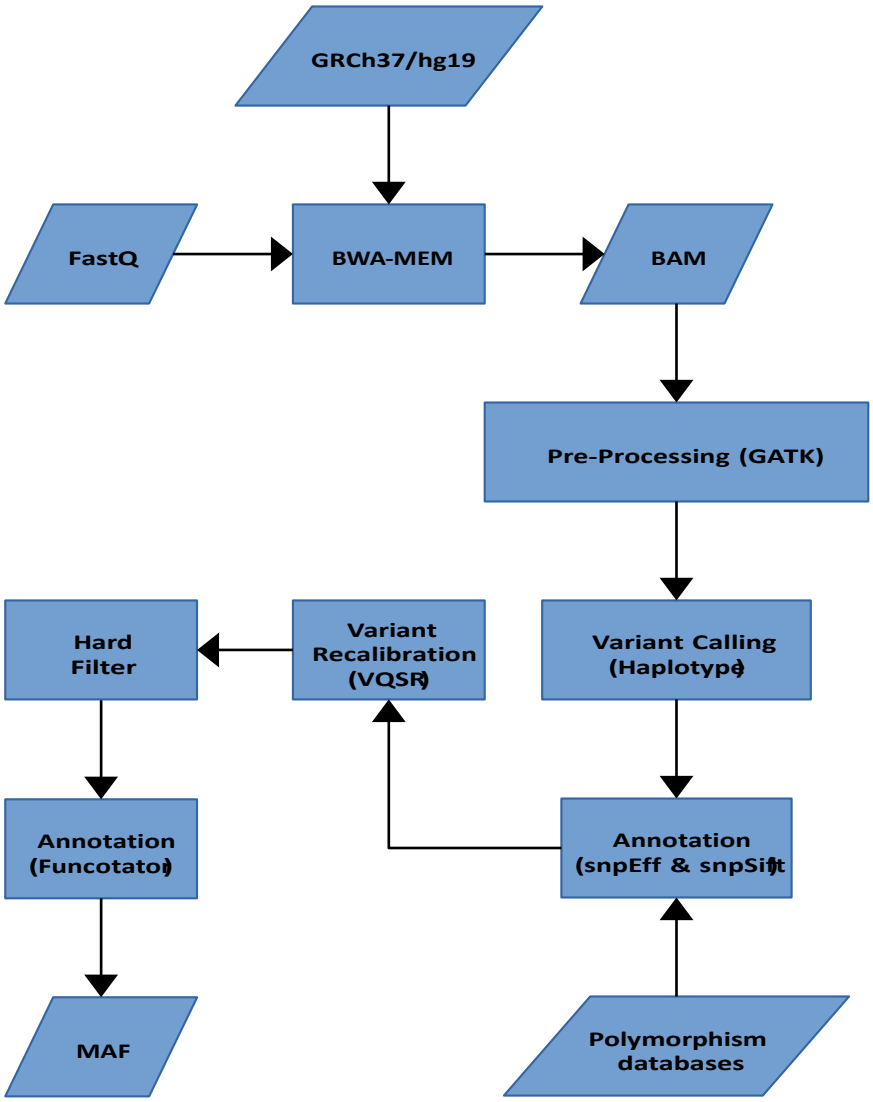
http://doi.org/10.47626/1516-4446-2024-3958
Candidate genes related to spiritual mediumship: a whole-exome sequencing analysis of highly gifted mediums – Gattaz WF et al.

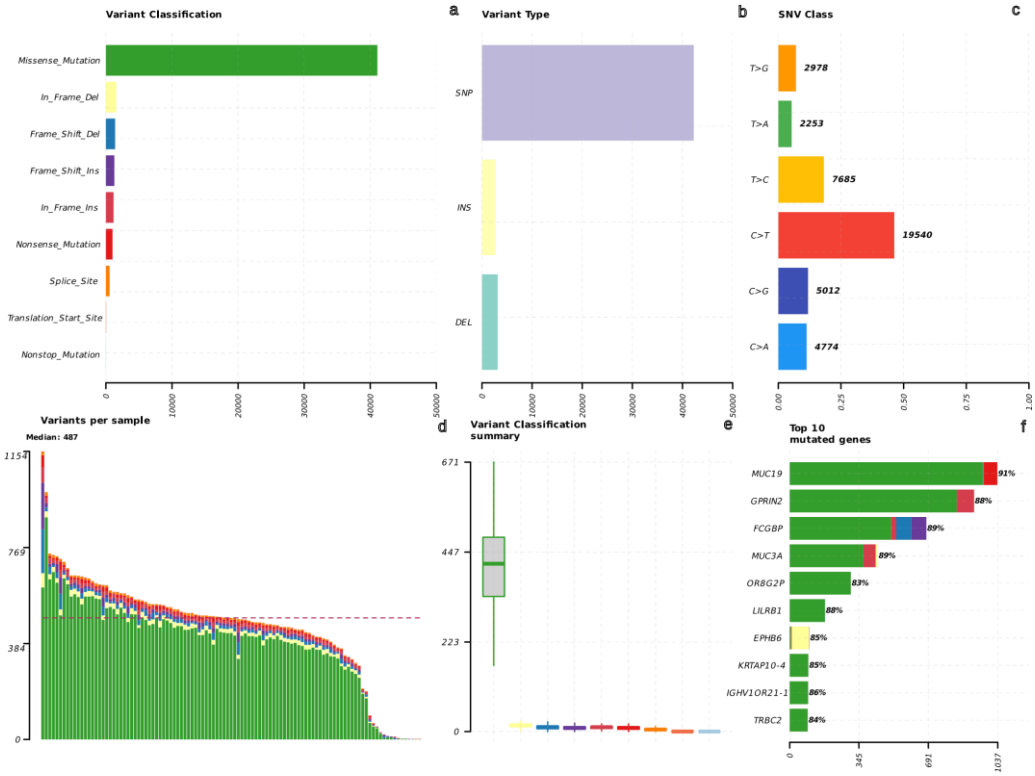
Supplementary Figure S1 Key steps of the pipeline utilized for processing exome data to produce a MAF file



Initially, sequencing reads (FastQ files) are aligned to the human reference genome (GRCh37/hg19) using BWA-MEM (v0.7.17-r1188), adjusting the thread count to 8. Subsequently, aligned files (BAMs) undergo preprocessing with GATK version 4.2, including steps like MarkDuplicates, BaseRecalibrator, and PrintReads with default parameters and 8 threads. Variant calling is conducted using HaplotypeCaller with specific parameters for each sample, followed by CombineGVCs and GenotypeGVCFs to identify variants in a “joint analysis” format. Identified variants are annotated using snpEff and SnpSift software, leveraging various DNA polymorphism databases. Filtering steps include GATK's Variant Recalibration (VQSR) method with specific parameters and a hard filter based on GATK Best Practices. Common variants (>1% variant allele frequency [VAF]) found in non-M subjects, potentially less correlated with observed phenotypes, are removed after comparison with public databases including dbSNP, 1000 Genomes, gnomAD, and ABRaOM. Finally, Funcotator annotates the remaining variants and the MAF file is generated.

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Supplementary Figure S2 Summary of the distribution of non-synonymous variants with depth > 8 and VAF > 30%



A) Classification of variants by type of alteration, with the number of variants displayed on the x-axis. B) Types of variants. C) SNV class that shows the most frequent nucleotide substitutions. D) Distributions of the variants per sample, with the dashed line representing the median (487 variants per sample). E) Box plot of the classification of variants. F) Top 10 genes with the highest number of variants on the x-axis. The color code of panels D, E, and F corresponds to the variant classification. The percentages of participants that have at least one variant of these genes are displayed on the right side of the bars. DEL = deletion; INS = insertion; SNP = single nucleotide polymorphism; SNV = single nucleotide variant.

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Supplementary Table S3 Genes in which mutations were detected in at least one-third of mediums in this study

Gene name	Tissue with highest expression[†]	Function	Frequency, n (%)				
			Mediums sample (n=54)	Twins[‡] (n=2)	Validation sample[§] (n=12)	Sensory system	Pineal gland[‖]
Mucin 19	Minor salivary gland	Mucous protection of epithelial tissues	47 (87.04)	2 (100)	11 (91.67)	-	X
Mucin 3a	Small intestine	Mucous protection of epithelial tissues	36 (66.67)	2 (100)	11 (91.67)	-	-
Mucin 4	Colon	Mucous protection of epithelial tissues	35 (64.81)	2 (100)	8 (66.67)	-	-
Major histocompatibility complex class II DR Beta 5	Lung	Antigen presentation	34 (62.96)	2 (100)	9 (75)	-	-
Zinc finger protein 717	Thyroid	Transcriptional regulation	31 (57.41)	1 (50)	8 (66.67)	-	-
Major histocompatibility complex class II DQ Beta 1	Lung	Antigen presentation	31 (57.41)	2 (100)	8 (66.67)	-	-
Major histocompatibility complex class II DR Beta 1	Lung	Antigen presentation	30 (55.56)	2 (100)	9 (75)	-	-
Transmembrane phosphatase with Tensin homology	Testis	Signal transduction	28 (51,85)	2 (100)	11 (91.67)	-	-
Notch receptor 4	Adipocytes	Regulates cell fate determination	27 (50.00)	2 (100)	5 (41.67)	-	-
Ephrin type-B receptor 6	Brain cortex	Modulates cell adhesion and migration	27 (50.00)	2 (100)	10 (83.33)	-	-
Fc gamma binding protein	Colon	Maintenance of mucosal structure	27 (50.00)	2 (100)	11 (91.67)	-	-

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Transmembrane Inner ear	Pituitary gland	Protein/vesicle trafficking	24 (44.44)	-	7 (58.33)	-	-
Anoctamin 7	Prostate gland	Cell-cell interaction	24 (44.44)	2 (100)	7 (58.33)	-	-
MAF bZIP transcription factor A	Skeletal muscle	Transcription factor that regulates pancreatic beta cell-specific expression.	23 (42.59)	-	8 (66.67)	-	-
Leukocyte immunoglobulin like receptor B3	WBC	Receptor of class I MHC antigens	23 (42.59)	-	7 (58.33)	-	-
Nuclear pore assoc. protein 1	Testis	Unknown (nucleoporin-derived?)	23 (42.59)	-	7 (58.33)	-	-
Sperm protein associated with the nucleus on the X chromosome	Testis	Regulates transcription and translation of several testis-specific genes	22 (40.74)	2 (100)	8 (66.67)	-	-
Protein phosphatase, Mg^{2+}/Mn^{2+} dependent 1E	Brain	Inactivates CaM kinases	22 (40.74)	2 (100)	9 (75)	-	-
LOC101930307	Unknown	Unknown	22 (40.74)	-	11 (91.67)	-	-
Olfactory receptor Family 8 Subfamily G member 2	Testis	Olfactory receptor	21 (38.89)	-	9 (75)	-	-
Immunoglobulin heavy constant gamma P (non-functional)	Lymph node	Pseudogene	21 (38.89)	2 (100)	6 (50)	-	-
Myosin phosphatase Rho interacting protein	Prostate gland	Regulates actin cytoskeleton	21 (37.04)	-	6 (50)	-	-
Mitochondrial ribosomal protein L45	Breast	Pseudogene	20 (37.04)	-	8 (66.67)	-	-
Putative POM121-like	Testis	Unknown	20 (37.04)	-	6 (50.00)	-	-

Immunoglobulin heavy variable 3-20	Lymph node	Antigen recognition	19 (35.19)	-	11 (91.67)	-	-
Leukocyte immunoglobulin like receptor B1	Spleen	Receptor of class I MHC antigens	19 (35.19)	-	11 (91.67)	-	-
DDHD domain containing 1	Testis	Phospholipase A1	19 (35.19)	-	8 (66.67)	-	-
Cytochrome P450 family 2 subfamily D member 6	Liver	Metabolism of fatty acids, steroids, and retinoids	19 (35.19)	-	9 (75)	-	-
Ring finger protein 212	Testis	Regulator of crossing-over during meiosis	19 (35.19)	-	9 (75)	-	-
Olfactory receptor family 5 subfamily H member 6	Testis	Olfactory receptor	19 (35.19)	2 (100)	10 (83.33)	hsa04740	-
G-protein regulated inducer of neurite outgrowth 2	Skin	Neurite outgrowth	18 (33.33)	-	11 (91.67)	-	-
Glycophorin E	Spleen	Minor sialoglycoprotein of erythrocyte membranes	18 (33.33)	-	5 (41.67)	-	-
Olfactory receptor family 10 subfamily G member 2	Liver	Olfactory receptor	18 (33.33)	-	9 (75)	hsa04740	-

CNS = central nervous system; RNA = ribonucleic acid; WBC = white blood cells.
Gene name, tissue expression, and function are given as provided by genecards.org.
† According to RNA-Seq data from genecards.org.
‡ A pair of monozygotic twins exhibiting mediumship.
§ Twelve mediums without controls used for independent validation.
‖ X indicates that the corresponding gene is on the gene set related to pineal gland function.