

Table 1. Summary of human variants reported in MYO5A with clinical significance annotations related to Griscelli Syndrome type 1.

SNP ID	Clinical significance and condition	Chr15pos	Variation	Type	PMID	References
rs764371254	Pathogenic: Griscelli syndrome, Type 1	52,376,435(-)	Ref/Alt: G/A NM_000259.3(MYO5A):c.2332C>T (p.Arg778Ter)	nonsense	9207796 12058346	Pastural <i>et al.</i> , 1997 Anikster <i>et al.</i> , 2002
rs2141121140	Pathogenic: Griscelli syndrome, Type 1	52,379,723(-)	Ref/Alt: G/A NM_000259.3(MYO5A):c.2110C>T (p.Gln704Ter)	nonsense	32275080	Elmaksound <i>et al.</i> , 2020
rs2043479059	Pathogenic: Griscelli syndrome, Type 1	52,416,294(-)	Ref/Alt: G/A NM_000259.3(MYO5A):c.463C>T (p.Arg155Ter)	nonsense	32275080	Elmaksound <i>et al.</i> , 2020
rs769021352	Pathogenic: Griscelli syndrome, Type 1	52,383,090(-)	Ref/Alt: C/A NM_000259.3(MYO5A):c.2012+1G>T	splice donor	25326635	Yang <i>et al.</i> , 2014
14070*	Pathogenic: Griscelli syndrome, Type 1	52,307,283(-)	47-BP INS, NT4634	insertion	10704277	Pastural <i>et al.</i> , 2000
rs2141084327	Likely pathogenic: Griscelli syndrome, Type 1	52,370,192(-)	Ref/Alt: G/A NM_000259.3(MYO5A):c.3043C>T (p.Arg1015Ter)	nonsense	-	CENTOGENE, 2021+
rs201961375	Uncertain significance: Griscelli syndrome, Type 1	52,330,351(-)	Ref/Alt: G/A NM_000259.3(MYO5A):c.4480+2C>T	splice donor	17576681	Buratti <i>et al.</i> , 2007
rs1446850592	Uncertain significance: Griscelli syndrome, Type 1	52,410,433(-)	Ref/Alt: C/T NM_000259.3(MYO5A):c.656G>A (p.Arg219His)	missense	-	KMC-MAHE, 2022 +
rs2041830255	Uncertain significance: Griscelli syndrome, Type 1	52,383,173(-)	Ref/Alt: G/A NM_000259.3(MYO5A):c.1930C>T (p.His644Tyr)	missense	-	Revvity Omics, 2024 +
rs374266181	Uncertain significance: Griscelli syndrome, Type 1	52,375,354(-)	Ref/Alt: C/T NM_000259.3(MYO5A):c.2527G>A (p.Val843Ile)	missense	-	Revvity Omics, 2024 +
2627586*	Uncertain significance: Griscelli syndrome, Type 1	52,383,145(-)	Ref/Alt: G/A NM_000259.3(MYO5A):c.1958C>T (p.Thr653Ile)	missense	-	IHG-UKL, 2023 +
rs561485938	Uncertain significance: Griscelli syndrome, Type 1	52,375,443(-)	Ref/Alt: C/T NM_000259.3(MYO5A):c.2438G>A (p.Arg813His)	missense	-	IIGK-IIG, 2024 +
rs756082685	Uncertain significance: Griscelli syndrome, Type 1	52,319,260(-)	Ref/Alt: G/C NM_000259.3(MYO5A):c.4959C>G (p.Ile1653Met)	missense	-	IIGK-IIG, 2024 +
rs751411629	Uncertain significance: Griscelli syndrome, Type 1; Inborn genetic diseases	52,367,093(-)	Ref/Alt: A/C NM_000259.3(MYO5A):c.3098T>G (p.Leu1033Trp)	missense	- -	BMGL-KAMC, 2018 + Ambry Genetics, 2024 +
rs752461164	Uncertain significance: Griscelli syndrome, Type 1 Likely benign: MYO5A-related disorder	52,375,404(-)	Ref/Alt: C/T NM_000259.3(MYO5A):c.2477G>A (p.Arg826His)	missense	- -	Baylor Genetics, 2021 + Prevention Genetics, 2024 +
rs72734962	Likely benign: Griscelli syndrome, Type 1 Benign: Inborn genetic diseases	52,317,044(-)	Ref/Alt: C/T NM_000259.3(MYO5A):c.5334+4G>A	intron	- -	CG-ARHLCHC, 2023 + Ambry Genetics, 2024 +
rs748268948	Likely benign: Griscelli syndrome, Type 1	52,416,223(-)	Ref/Alt: T/G NM_000259.3(MYO5A):c.534A>C (p.Arg178=)	synonymous	-	CG-ARHLCHC, 2023 +
rs56132571	Likely benign: Griscelli Syndrome, Type 1; Intellectual disability; Inborn genetic diseases	52,367,055(-)	Ref/Alt: C/T NM_000259.3(MYO5A):c.3136G>A (p.Val1046Met)	missense	- - -	AJCGC-AJCSH, 2021 + CBPG-CHRUL, 2020 + Ambry Genetics, 2024 +
rs1058219	Benign: Griscelli syndrome, Type 1	52,351,367(-)	Ref/Alt: G/A NM_000259.3(MYO5A):c.3736C>T (p.Arg1246Cys)	missense	9207796 10733681	Pastural <i>et al.</i> , 1997 Lambert <i>et al.</i> , 2000
rs4774621	Benign: Griscelli syndrome, Type 1	52,383,064(-)	Ref/Alt: T/C NM_000259.3(MYO5A):c.2012+27A>G	intron	-	Genome Nilou, 2021 +
rs11386962	Benign: Griscelli syndrome, Type 1	52,348,793(-)	Ref/Alt: T/TA NM_000259.3(MYO5A):c.3858+12dup	intron	-	Genome Nilou, 2021 +
rs72623980	Benign: Griscelli syndrome, Type 1	52,410,502(-)	Ref/Alt: T/C NM_000259.3(MYO5A):c.613-26A>G	intron	-	Genome Nilou, 2021 +
rs2414145	Benign: Griscelli syndrome, Type 1	52,375,355(-)	Ref/Alt: G/A NM_000259.3(MYO5A):c.2526= (p.Ile842=)	no sequence alteration	-	Genome Nilou, 2021 +
rs1724577	Benign: Griscelli syndrome, Type 1	52,397,434(-)	Ref/Alt: T/G NM_000259.3(MYO5A):c.1086= (p.Asp362=)	no sequence alteration	-	Genome Nilou, 2021 +
rs1724623	Benign: Griscelli syndrome, Type 1	52,389,376(-)	Ref/Alt: A/G NM_000259.3(MYO5A):c.1543-13T>C	intron	-	Genome Nilou, 2021 +
rs61363656	Benign: Griscelli syndrome, Type 1	52,416,305(-)	Ref/Alt: T/C NM_000259.3(MYO5A):c.456-4A>G	intron	-	Genome Nilou, 2021 +
rs71425744	Benign: Griscelli syndrome, Type 1	52,389,447(-)	Ref/Alt: AT/A NM_000259.3(MYO5A):c.1543-84del	intron	-	Mendelics, 2019 +
rs2290333	Benign: Griscelli syndrome, Type 1	52,319,274(-)	Ref/Alt: G/T NM_000259.3(MYO5A):c.4945C>A (p.Arg1674=)	synonymous	-	Fulgent Genetics, 2022 +

Genomic location annotation version GRCh38; Variants information is currently extracted from OMIM, ClinVar VCF, dbSNP and UniProt's Human polymorphisms and disease mutations database. * ClinVar Accession; + ClinVar Submitter.