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SHORT COMMUNICATION

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Survey of functional Mendelian variants in New Zealand Huntaway and Heading dog breeds

Abstract

New Zealand (NZ) Huntaway and Heading dogs are working breeds that play active roles on farms across NZ. While these breeds are common in NZ, they are not well-known elsewhere, and little is understood about their genetic make-up. Here, we used whole genome sequencing to provide a comprehensive genomic view of 249 working dogs. As first use of this resource, we report the allele frequencies of provisionally functional variants aggregated from the Online Mendelian Inheritance in Animals (OMIA) database. Of 435 "probably causal" variants, 27 segregated in our sample. Notable examples of disease variants potentially actionable for selection include those in the *CUBN*, *CLN8*, *SGSH*, *SOD1*, *VWF*, and *VPS13B* genes. These findings will enable genetic testing and selection opportunities to help improve the health and performance of future generations of these unique breeds.

Agriculture is New Zealand (NZ)'s largest industry and livestock farming relies on the ~200 000 farm dogs working across the country (Isaksen et al., 2020). Despite Huntaway and Heading dog breeds representing most of these dogs, no large-scale genetic studies have yet been conducted on them. Figure 1 illustrates individuals characteristic of the two breeds, although substantial interindividual variation exists in morphology and appearance. Most Huntaways are large (average weight = 28 kg) and are selected for their loud bark and endurance, allowing them to drive livestock from behind at large distances (Cave et al., 2009). Descended from Border Collies, Heading dogs are of medium body size (average weight = 19 kg) and are discouraged from barking. Instead, they are selected to stare down livestock in close quarters (Cave et al., 2009).

The genetic variation and prevalence of most diseases is understudied in Huntaways and Heading dogs. The Online Mendelian Inheritance in Animals (OMIA) database (Nicholas et al., 1995) provides a catalogue of trait-linked variants (predominantly Mendelian in effect) in dogs, detailing the majority of loci targeted for diagnostic testing and parent selection for breeding companion breeds. The genotype status of these loci is unknown in NZ farm dogs, meaning that the most relevant variants and which tests might benefit these unique breeds are unknown.

Here, we aimed to survey functional Mendelian variants from OMIA in NZ Huntaways and Heading dogs. To this end, we collected blood samples from 249 dogs for whole genome sequencing. This sample included 130 Huntaways, 104 Heading dogs, and 15 Huntaway/Heading intercrosses or crosses of other working breeds, with breed declared by dog owners. Sampling of relatives was avoided where known, and animals were sourced across several NZ regions (Table S1).

DNA was extracted from 200 μL of whole blood using a Qiagen MagAttract HMW DNA Kit (Cat67563 Qiagen NZ), and sequencing libraries were produced from 350 ng of DNA using an Illumina DNA Prep Kit (Illumina, NZ). Sequencing was performed on an Illumina NovaSeq 6000 using S4 300 flowcells (2 × 150-bp reads; conducted at GeneMark, Hamilton, NZ).

Read mapping was performed using BWA-MEM (v0.7.17) (Jung & Han, 2022) referencing the canFam4 (UU_Cfam_GSD_1.0) genome (Wang et al., 2021), yielding BAM files with an average read depth of 22.88x and average read quality of 35.68. Per-sample variant calling was performed with GATK HaplotypeCaller (4.5.0) (Depristo et al., 2011), intermediate GVCF files were merged across samples with GenomicsDBImport and joint genotyping was performed with GenotypeGVCFs (N = 20 060 548 variants). Quality filtering included application of GATK-recommended hard filters and additional genotype quality thresholds (per-sample DP > 8 and GQ > 20). These criteria yielded 16 678 350 variants (12 059 693 single nucleotide variants [SNVs] and 4 618 657 indels).

The positions of 552 variants were retrieved from OMIA, including all variants reported as "probably causal" in dogs as of March 2025. Where alternative genomes were referenced, 100-bp flanking sequences were extracted using the faidx method from SAMtools

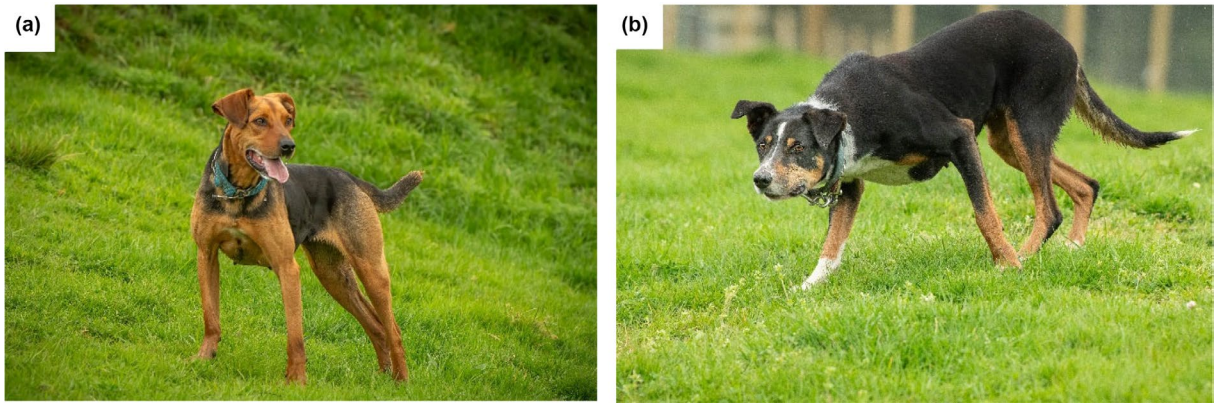

FIGURE 1 Example images of New Zealand farm dog breeds. (a) Huntaway. (b) Heading dog.

(v1.16.1) and remapped to canFam4 (Wang et al., 2021) using BWA-MEM (v0.7.17). These loci were intersected with the population VCF file using BCFtools (1.19) (Danecek et al., 2021) to identify those segregating in at least one dog in the sample. Variant positions and quality were verified through visual inspection of genome alignments in carrier animals using Integrative Genome Viewer software (Robinson et al., 2011). Given the difficulty in systematically characterising large structural variants from short read sequence data, this category of variant was omitted from our analysis ($N = 108$ variants). Nine variants were excluded due to having no reported positions.

Query of the genome sequence dataset identified 27 loci that were polymorphic in our sample; allele frequencies are described in Table S2. Of these variants, 11 were previously reported to impact morphological/aesthetic traits (e.g. coat colour/patterning), whilst 16 were previously implicated in disease. Of the 435 variant loci queried, 408 did not segregate in our sample and are listed in Table S3. Given that dogs were selected to represent multiple geographic regions, the invariability of these positions suggests their frequencies are either low or absent from Huntaway and/or Heading dog breeds.

The OMIA database represents variant discoveries with evolving evidence of causality (Nicholas et al., 1995). On this basis, we subjectively evaluated the literature to assess the functional candidacy of each variant, aiming to highlight the most compelling variants for use as selection diagnostics. We identified six variants in the *CUBN*, *CLN8*, *SGSH*, *SOD1*, *VWF*, and *VPS13B* genes that fulfil these criteria. Table 1 lists these variants, and a brief discussion of their molecular and clinical impacts follows.

A 1-bp deletion that causes a loss of function (LOF) in the *CUBN* (cubilin) gene was heterozygous in six dogs in our sample (all Heading dogs), equating to a minor allele frequency (MAF) of 2.9%. Cubilin is essential for cobalamin (vitamin B12) uptake (Owczarek-Lipska et al., 2013). The deletion was discovered in Border Collies in 2013 and leads to potentially life-threatening malabsorption of the vitamin when left untreated (Owczarek-Lipska et al., 2013). Previously reported allele frequencies varied between breeds, but have been reported as lower (1.5%) in Heading dogs' closest known relatives, Border Collies. (Mizukami et al., 2016). We identified carriers in Southland (four), Waikato (one), and Hawkes Bay (one).

A recessive allele in the *CLN8* gene (ceroid-lipofuscinosis, neuronal 8), first reported in 2014 in an Australian Shepherd/Blue Heeler cross, leads to a severe neurodegenerative disorder called neuronal ceroid lipofuscinosis (Guo et al., 2014). Neuronal ceroid lipofuscinosis is characterised by loss of motor functions, seizures, and blindness (Guo et al., 2014). This disease has no treatment and affected dogs are usually euthanised early in life. We identified 21 carriers of this nonsense SNV (13 Heading dogs, seven Huntaways, and one Huntaway/Heading dog cross) in multiple regions (nine in Southland, six in Hawkes Bay, four in Waikato, and two in other regions). The MAF is 6.3% and 2.7% in Heading dogs and Huntaways respectively. Like the *CUBN* variant, this frequency is considerably higher than reported in the breed of discovery. Previous studies profiling Blue Heelers, Australian Shepherds, and German Shorthaired Pointers showed allele frequencies of 0%, 0.67%, and 0% respectively (Guo et al., 2014, 2019). Although we are unaware of affected dogs presenting in NZ, these can be assumed to exist in appreciable numbers, highlighting the *CLN8* variant as an obvious candidate for future testing.

A 1-bp insertion in *SGSH* (heparan sulfate sulfamidase) causes a deficiency of this enzyme and leads to a recessive, untreatable neurodegenerative disorder called mucopolysaccharidosis (MPS) IIIA (Yogalingam et al., 2002). Discovered in Huntaways in 2002 from one of the few previous genetic studies of the breed, this LOF allele causes severe MPS. By contrast, Dachshunds exhibit a less severe MPS presentation caused by an alternative, partial LOF allele (Aronovich et al., 2000). In Huntaways, onset begins from age 18 months, with progressive symptoms that resemble cerebellar disease (Yogalingam et al., 2002). The allele frequency of

TABLE 1 Summary of candidate Mendelian variants for selection in NZ farm dogs.

OMIA ID [a]	Gene	Chr[b]	Variant[b]	Phenotype[a]	Alternative allele frequency[b]			Genotype frequencies		
					Huntaways[c]	Heading dogs[c]	Total[d]	Huntaways[c]	Heading dogs[c]	Total[d]
447	*CUBN*	2	g.18932445delp. (Q2798Rfs*)	Intestinal cobalamin malabsorption[a]	0	0.029	0.012	C/C=1 C/−=0 −/−=0	C/C=0.942 C/−=0.057 −/−=0	C/C=0.975 C/−=0.024 −/−=0
338	*CLN8*	37	g.30769171G>A p.(W195*)	Neuronal ceroid lipofuscinosis	0.027	0.063	0.042	G/G=0.946 G/A=0.054 A/A=0	G/G=0.875 G/A=0.125 A/A=0	G/G=0.916 G/A=0.084 A/A=0
577	*SGSH*	9	g.2406797insA p.(Y229*)	Mucopolysaccharidosis IIIA	0.004	0	0.01	T/T=0.992 T/TA=0.008 TA/TA=0	T/T=1 T/TA=0 TA/TA=0	T/T=0.98 T/TA=0.02 TA/TA=0
36	*SOD1*	31	g.27123057G>A p.(E40K)	Degenerative myelopathy	0.215	0.019	0.124	G/G=0.608 G/A=0.354 A/A=0.038	G/G=0.962 G/A=0.038 A/A=0	G/G=0.771 G/A=0.209 A/A=0.02
401	*VWF*	27	g.7140281C>T p.(S2479S)	Von Willebrand disease I	0.035	0	0.022	C/C=0.931 C/T=0.069 T/T=0	C/C=1 C/T=0 T/T=0	C/C=0.96 C/T=0.036 T/T=0.004
478	*VPS13B*	13	g.1522613del4 p.(V595Ifs*)	Trapped neutrophil syndrome	0.023	0.01	0.016	TTGT/TTGT=0.954 TTGT/−=0.046 −/−=0	TTGT/TTGT=0.981 TTGT/−=0.019 −/−=0	TTGT/ TTGT=0.968 TTGT/−=0.032 −/−=0

Abbreviations: Chr, chromosome; del, deletion.

[a]Data were obtained from the OMIA database (3).

[b]Relative to the canFam4 reference genome assembly.

[c]Calculated from dogs reported as purebred.

[d]Calculated from total sample.

the insertion in Huntaways was estimated to be 3.8% in 2002. We observed five carriers in our sample; however, four were associated with a dog colony where the disease was previously studied. Removing these dogs, we observed an allele frequency of 0.4% in Huntaways (one carrier in Hawkes Bay). Although we report a low frequency, the severity of the disease suggests that routine genetic testing would be beneficial for early diagnosis and selective mating.

Carriers and homozygotes of a missense variant associated with degenerative myelopathy (Awano et al., 2009) were identified in our cohort. This SNV impacts SOD1 (superoxide dismutase 1), an enzyme that breaks down superoxide radicals. Degenerative myelopathy is an adult-onset degeneration of the spinal cord that causes paraplegia and is a model for amyotrophic lateral sclerosis (Awano et al., 2009). It is untreatable and affected dogs are often euthanised. We identified 52 carriers (46 Huntaways, four Heading dogs, and two Huntaway crosses) and five homozygotes (all Huntaways) across NZ (23 in Hawkes Bay, 16 in Southland, eight in Waikato, five in Otago, and five in other regions). The allele frequency in Huntaways (21.5%) is high but comparable to that reported in German Shepherds (22%), while the frequency in Heading dogs (1.9%) is low compared to other breeds (Maki et al., 2022; Santos et al., 2020). The allele exhibits variable penetrance, with some breeds presenting high frequencies yet no affected dogs (Zeng et al., 2014). The disease also has a late onset, meaning homozygotes may develop symptoms later in life or not at all (Awano et al., 2009). Given the high frequency amongst Huntaways, further testing would be beneficial to help assess risk in this breed.

A splice site variant in *vWF* (von Willebrand factor) was identified in our sample and has been proposed to cause the bleeding disorder type 1 von Willebrand disease (vWD) in several breeds (Crespi et al., 2018; Donner et al., 2016; Gentilini & Turba, 2013). First described in 2004, this is the least severe form of vWD and results in a reduced concentration of vWF, a glycoprotein required for blood clotting (Venta et al., 2014). We observed nine carriers (all Huntaways) and one homozygote (Huntaway cross) in several regions (three in Waikato, three in Hawkes Bay, and four in other regions). This equates to a MAF of 2.2% in Huntaways. Literature suggests that this allele exhibits incomplete penetrance with ~40% of homozygotes in previous studies developing the disease (Crespi et al., 2018). Since the physical demands of work mean that injuries are common amongst farm dogs, and vWD increases risk of prolonged bleeding, testing to assess the severity of risk in Huntaways and avoidance of carrier-carrier mating would be beneficial (Isaksen et al., 2020).

A 4-bp deletion in *VPS13B* was detected at an allele frequency of 2.3% in Huntaways (six carriers) and 1% in Heading Dogs (two carriers), with carriers in Southland and Hawkes Bay. First discovered in Border Collies in 2011, this allele causes trapped neutrophil syndrome (TNS), a deficiency of segmented neutrophils that leads to a compromised immune system, pyrexia, and lameness (Shearman & Wilton, 2011). While some treatments can prolong life, affected dogs usually die or are euthanised young (Suciu et al., 2024). Variants in *VPS13B* have been shown to cause Cohon syndrome in humans, which is homologous to TNS (Shearman & Wilton, 2011). TNS was first described in a population of Australian and NZ Border Collies. It is therefore unsurprising that the causal variant was detected in a closely related, and probably crossbred, population. We observed a low allele frequency compared to populations of Border Collies in Japan (7%) and Norway (8%). However, the severity of TNS makes this variant a strong candidate for testing and selection.

While variants impacting morphological/aesthetic traits were detected in our sample, the current study focused foremost on variants known or assumed to cause disease. One variant that potentially sits in both those categories, however, is a splice variant in *MLPH*. This allele causes recessive coat colour dilution and has been suggested to predispose some breeds to black-hair follicular dysplasia (Drögemüller et al., 2007; Welle et al., 2009). It is notable therefore that black-hair follicular dysplasia has been previously reported in Huntaways (Munday et al., 2009), although the extent to which the *MLPH* dilution allele influences disease presentation in this breed is unknown. For the dilution phenotype at least, we could confirm this effect given that two of the four homozygous dogs in our sample for which photographs were available both had blue/grey coats.

We detected indels in *BTBD17* and *KCNJ10* with MAFs of 16% and 8% respectively (Table S2). The variant in *BTBD17* was associated with embryonic lethality in German Shorthaired Pointers but its causality has been questioned due to high frequencies in other breeds (Meadows et al., 2023; Meyers-Wallen et al., 2017). The *KCNJ10* variant has been associated with severe ataxia in terriers with weak evidence of causality (Gast et al., 2016). The high frequencies observed in our sample likewise suggest that these alleles are unlikely to be causal in the NZ population.

In conclusion, we present a comprehensive genetic survey of previously published functional Mendelian variants in Huntaways and Heading dogs. We highlight six diagnostic candidates that would benefit these breeds, where further testing, genotyping, and/or mate selection would help assess breed-specific risk, manage allele frequencies, and avoid genetic disease.

KEYWORDS
bioinformatics, *Canis lupus familiaris*, Heading dog, Huntaway, Mendelian genetics, whole genome sequencing

AUTHOR CONTRIBUTIONS

Florence Smith: Formal analysis; investigation; writing – original draft; writing – review and editing. **Thomas Lopdell:** Data curation; formal analysis; software; supervision; writing – review and editing. **Melissa Stephen:** Conceptualization; data curation; investigation; resources. **Millicent Henry:** Investigation; formal analysis. **Keren Dittmer:** Investigation; writing – review and editing; resources. **Hayley Hunt:** Investigation; writing – review and editing; resources. **Nick Sneddon:** Investigation; writing – review and editing; project administration; resources. **Liam Williams:** Investigation; resources. **Jack Rolfe:** Investigation; resources. **Dorian Garrick:** Investigation; writing – review and editing; funding acquisition. **Mathew D. Littlejohn:** Conceptualization; funding acquisition; project administration; supervision; validation; writing – review and editing; resources.

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CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

ETHICS STATEMENT

All experiments were performed in strict accordance with the rules and guidelines outlined in the New Zealand Animal Welfare Act 1999. Samples were gathered in accordance with protocols approved by the Massey University Animal Ethics Committee, Palmerston North, New Zealand (approval MUAEC 23/37). No animals were sacrificed for this study.

DATA AVAILABILITY STATEMENT

The sequence data were submitted to the NCBI Sequence Read Archive (accession no. PRJNA1301006), and are accessible through the following link (https://www.ncbi.nlm.nih.gov/sra/PRJNA1301006).

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.