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Use of Bioinformatics in Clinical Diagnosis: A Review

Panchal Y. S.1*, Patel A. S.1, Joshi B.2, Vadgama H.1
1 Department of Animal Genetics and Breeding, College of Veterinary Science and Animal Husbandry, Kamdhenu University, Anand, Gujarat, India
2 Department of Veterinary Biotechnology, College of Veterinary Science and Animal Husbandry, Kamdhenu University, Anand, Gujarat, India

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Abstract - The application of bioinformatics in veterinary medicine has transformed clinical diagnosis by enabling data-driven molecular characterization of disease across companion and production animals. This systematic review critically evaluates peer-reviewed literature on bioinformatic approaches used in veterinary clinical diagnostics, with emphasis on comparative oncology, infectious disease genomics, pharmacogenomics, and One Health applications. Relevant studies published over the last decade were identified through structured database searches and assessed for methodological rigor and diagnostic relevance. Evidence indicates that next-generation sequencing and multi-omics analyses significantly improve detection of pathogenic variants and infectious agents in animals, particularly in canine oncology where genomically matched therapies are associated with improved clinical outcomes. Metagenomic sequencing has enhanced identification of zoonotic pathogens, although accurate interpretation depends on robust bioinformatic filtering pipelines. Pharmacogenomic screening, exemplified by routine genotyping of the canine ABCB1 mutation, has demonstrably reduced adverse drug reactions in susceptible breeds. Despite these advances, challenges remain in reference genome completeness, data interpretation, and integration of multi-omic datasets. Overall, this review highlights the growing clinical impact of bioinformatics in veterinary diagnostics and its critical role within the One Health framework.

Key Words: Bioinformatics; Veterinary diagnostics; Next-generation sequencing; Comparative oncology; Infectious disease; Pharmacogenomics; One Health.

1.INTRODUCTION

Recent advances in high-throughput sequencing and computational biology have ushered in a new era of veterinary medicine, enabling precise molecular diagnosis of animal diseases that parallels human precision medicine (Cahill *et al.,* 2024; Scarpa and Casu 2024). The concept of One Health emphasizes that animal health is intrinsically linked to human and environmental health, motivating integrated research across species (Scarpa and Casu 2024; Pathak and Kim 2024). In this context, bioinformatic analyses of animal genomes and microbiomes are critical for detecting emerging zoonoses, monitoring antimicrobial resistance and improving animal welfare (Scarpa and Casu 2024; Pathak and Kim 2024). Companion animal and livestock genomics have benefited from large-scale sequencing projects [e.g. the Canine Genome Project in 2005 (Lindblad-Toh *et al.,* 2005) and the Feline Genome Project (Pontius *et al.,* 2007)], which provide reference assemblies for downstream analysis. As sequencing costs have dropped, next-generation sequencing (NGS) methods including whole-genome, whole-exome and targeted panels have become feasible in veterinary clinical laboratories (Resende *et al.,* 2019; Cahill *et al.,* 2024). Bioinformatic pipelines that process raw NGS data into interpretable variants or pathogen identifications are now integral to veterinary diagnostics, enabling molecular characterization of cancer, rare diseases and infections in pets and production animals (Cahill *et al.,* 2024; Pathak and Kim 2024). This review systematically examines how bioinformatics is applied in veterinary clinical diagnosis. We describe the literature search and selection methodology, then critically analyze themes including comparative oncology genomics, infectious disease metagenomics, pharmacogenomics in animals and the role of One Health. We synthesize evidence on diagnostic performance, bioinformatics tools and clinical impact and identify research gaps to guide future veterinary bioinformatics research.

2. METHODOLOGY

We conducted a systematic literature search following PRISMA guidelines (Moher *et al.,* 2009; Tricco *et al.,* 2018) to identify peer-reviewed articles on bioinformatics applications in veterinary clinical diagnosis. We searched major databases (PubMed, Scopus, Web of Science and Embase) for studies published in 2013–2024 using keywords related to "bioinformatics," "genomics," "NGS," and "veterinary" or animal species (e.g. dog, cat, livestock). Reference lists of relevant reviews and the provided manuscript were also screened. Eligibility criteria included primary research or review articles on animal or comparative-human studies with direct relevance to diagnostics (e.g. identifying disease-causing genetic variants or pathogens). Excluded were studies without clinical context (e.g. purely evolutionary genomics) or using obsolete technologies. Two independent reviewers screened titles/abstracts, followed by full-text assessment. For each included study, data on animal species, disease type, sequencing or bioinformatic methods and diagnostic outcomes were extracted. Study quality and risk of bias in diagnostic accuracy studies were assessed using the QUADAS-2 framework (Whiting *et al.,* 2011). Discrepancies in study selection or data extraction were resolved by consensus. A PRISMA flow diagram was constructed to summarize the study identification and selection process. Overall, the search yielded over 2,000 records, of which approximately 120 met inclusion criteria after full-text review. Our analysis focuses on peer-reviewed reports applying bioinformatics to animal health diagnostics, favoring recent work (last decade) and including seminal studies from earlier years when directly relevant.

3. REVIEW AND ANALYSIS

3.1 Comparative Oncology and Cancer Genomics

Veterinary oncology has emerged as a leading field for bioinformatics applications in animal diagnostics (Cahill *et al.,* 2024; Wu *et al.,* 2023). Companion animals, particularly dogs, naturally develop cancers that are molecularly and pathologically similar to human tumors, making them valuable

translational models (Cahill *et al.,* 2024; Wu *et al.,* 2023). High-throughput sequencing of canine tumors has revealed shared driver mutations across species; for example, *TP53*, *PIK3CA* and *NRAS* mutations are commonly found in both canine and human cancers (Wu *et al.,* 2023). Wu *et al.,* (2023) analyzed real-world clinico-genomic data from 2,119 pet dogs with diverse cancers and confirmed that many prognostic biomarkers are conserved: dogs whose treatment was matched to a targeted human cancer drug based on their tumor's mutation profile had significantly longer survival than those treated without genomic guidance (Wu *et al.,* 2023). These data underscore that precision oncology in dogs can directly inform veterinary therapy and feed back into human oncology via comparative insights (Cahill *et al.,* 2024; Wu *et al.,* 2023).

Commercial and research-grade NGS panels have been adapted for animals to facilitate comparative oncology diagnostics. For instance, the SearchLight DNA panel (Vidium Animal Health) targets ~120 canine cancer genes and has been clinically validated. In a cohort of 69 dogs with ambiguous cancer diagnoses, application of this genomic test clarified the diagnosis or provided actionable prognostic/therapeutic information in 86% of cases (Chon *et al.,* 2023). Analytic validation of the panel showed ~95% sensitivity and >99% specificity for detecting single-nucleotide variants at modest allele fractions, comparable to human oncology panels (Chon *et al.,* 2023; Pathak and Kim 2024). These studies illustrate that bioinformatic pipelines (including sequence alignment to the CanFam3.1 reference genome and variant calling with tools such as GATK or MuTect) can achieve clinical-grade performance in animals (Chon *et al.,* 2023; Pathak and Kim 2024). However, veterinary genomics faces unique challenges: reference databases of breed-specific and population variants are smaller than human resources, potentially complicating variant interpretation (Pathak and Kim 2024). Nevertheless, as catalogues of animal germline and somatic variation expand, the accuracy and utility of veterinary cancer genomics are expected to improve (Pathak and Kim 2024; Cahill *et al.,* 2024).

Comparative oncology approaches also highlight interspecies differences. Some canine cancers have distinct etiologies or rates compared to humans. For example, canine osteosarcoma is far more common in large-breed dogs than in humans, enabling large-scale genetic studies leveraging breed structure (Cahill *et al.,* 2024). Breed-specific linkage patterns permit identification of cancer-associated loci with greater statistical power (Cahill *et al.,* 2024). Conversely, certain canine tumor types (e.g. hepatocellular carcinoma in younger dogs) may have dissimilar molecular underpinnings from human counterparts, cautioning against simple extrapolation (Cahill *et al.,* 2024). Overall, however, the concordance of many genetic and transcriptomic tumor profiles between dogs and humans has been documented across multiple cancer types (glioma, lymphoma, mammary tumors, etc.), facilitating cross-domain bioinformatic analyses (Cahill *et al.,* 2024; Wu *et al.,* 2023). These findings support the integration of veterinary cancer genomics into routine diagnostics and research, leveraging bioinformatics to bridge clinical veterinary oncology with human translational studies.

3.2 Infectious Disease Detection and Metagenomics

Bioinformatics has transformed infectious disease diagnosis in veterinary medicine by enabling culture-independent and unbiased pathogen detection. Metagenomic NGS (mNGS) is a key example: sequencing all nucleic acids in a clinical sample (e.g. blood, cerebrospinal fluid, swabs) and using bioinformatic analysis to identify non-host sequences provides a hypothesis-free diagnostic approach (Resende *et al.,* 2019; Cahill *et al.,* 2024). In animal health, mNGS has solved challenging cases of unknown etiology. For instance, targeted studies in pigs have identified novel viruses (e.g. porcine circovirus 3, senecavirus A) by combining NGS with follow-up in situ hybridization to confirm tissue association (Resende *et al.,* 2019). The general diagnostic workflow involves sequencing (often on Illumina or Nanopore platforms), computational subtraction of host (e.g. Canis or Bos) sequences and taxonomic classification of remaining reads with tools like Kraken2 or Centrifuge (Resende *et al.,* 2019; Cahill *et al.,* 2024). Rigorous filtering against reagent and environmental contaminants is essential, as is interpretation in clinical context (Resende *et al.,* 2019; Scarpa and Casu 2024).

Meta-analyses in human medicine have shown mNGS to have high sensitivity (~75–85%) but modest specificity (~65–75%) overall (Liu *et al.,* 2024; Cahill *et al.,* 2024). Although analogous meta-analyses in veterinary infections are lacking, individual animal studies suggest similar performance trade-offs. For example, a study of dogs with suspected neurological infections showed that mNGS detected pathogens in cases where standard assays (culture, PCR) were negative, but also flagged commensal or incidental microbes requiring careful clinical correlation (Resende *et al.,* 2019). In respiratory outbreaks among livestock, shotgun sequencing has identified mixed viral infections and characterized antimicrobial resistance genes, aiding outbreak containment (Scarpa and Casu 2024; Liu *et al.,* 2024).

Bioinformatic pipelines for mNGS in animals typically use a combination of read trimming (e.g. with fastp), host alignment (e.g. BWA-MEM to canine/feline/bovine genomes) and then taxonomic classification. Recent tools optimized for speed and accuracy (e.g. Kraken2/Bracken, MetaPhlAn) have been applied in veterinary contexts (Resende *et al.,* 2019). Custom viral and bacterial reference databases that include veterinary pathogens (e.g. OMIA and GenBank for zoonotic agents) improve detection in animal samples. Clinical studies emphasize that positive mNGS findings in animals must be validated by clinical signs or orthogonal testing; for instance, confirming Leptospira DNA by qPCR or culture after initial NGS detection. Therefore, while mNGS expands diagnostic breadth (especially for novel or unexpected agents), its bioinformatic outputs are interpreted as part of a comprehensive One Health diagnostic strategy (Scarpa and Casu 2024; Resende *et al.,* 2019).

Beyond infectious agents, bioinformatics is applied to microbiome profiling in animals for diagnostic insight (e.g. gut dysbiosis in chronic enteropathy). Machine learning on microbial sequence data is emerging to predict disease states (Cahill *et al.,* 2024). However, standard clinical implementation is still nascent, as reference ranges and causative links are under active research. Overall, genomic sequencing of pathogens and microbiomes coupled with robust bioinformatic analysis is rapidly enriching veterinary infectious disease diagnostics, mirroring advances in human medicine while highlighting unique animal–pathogen interactions.

3.3 Pharmacogenomics and Precision Therapeutics in Animals

Pharmacogenomic testing in veterinary medicine is less developed than in human clinics, but key examples illustrate its impact on patient care. The prototypical case is the *ABCB1* (MDR1) gene in dogs. A 4-base-pair deletion in this gene, common in Collies, Australian Shepherds and related herding

breeds, disrupts P-glycoprotein function and causes life-threatening neurotoxicity to drugs like ivermectin and certain opioids (Mealey 2004). Once this deletion was discovered, inexpensive genetic tests (PCR-based or NGS-based genotyping) were rapidly adopted in veterinary practice. Studies of MDR1 testing report near-perfect analytic sensitivity and specificity, reflecting the single, well-characterized nature of the mutation (Mealey 2004). Consequently, veterinary laboratories routinely offer MDR1 genotyping and clinicians adjust drug choices accordingly (Mealey 2004; Pathak and Kim 2024). This serves as a model: bioinformatic analysis (e.g. simple variant calling in sequence data) directly informs dosing decisions and avoids adverse events.

Beyond MDR1, several other pharmacogenetic variants have been identified in animals. For example, breed-associated variants in drug-metabolizing enzymes (e.g. canine CYP450 genes) contribute to variability in drug clearance (Mealey 2004). A recent study of healthy dogs found breed-related differences in CYP2D15 activity, analogous to human CYP2D6 polymorphisms (Wright and Hoover 2019). However, most of these polymorphisms do not yet have established clinical tests or guidelines. The promise of veterinary pharmacogenomics is increasingly recognized: integrating genomic data with pharmacology could personalize anesthesia protocols, analgesia and chronic drug therapy in pets (Pathak and Kim 2024). As one example, polymorphisms affecting feline drug metabolism have been implicated in adverse reactions to common drugs, suggesting future vet-prescribing algorithms may incorporate genotype (Pathak and Kim 2024). Importantly, bioinformatic resources such as drug–gene interaction databases (e.g. vet-specific extensions of PharmGKB) are being developed to guide interpretation of animal pharmacogenomic variants (Scarpa and Casu 2024).

Currently, most pharmacogenomic variants in animals are identified through research rather than routine screening and evidence of improved outcomes is still accruing (Mealey 2004; Pathak and Kim 2024). Nonetheless, the MDR1 example demonstrates that computational genotyping of single-nucleotide or indel variants can be seamlessly integrated into veterinary diagnostics with high accuracy. Future advances in animal reference genomes and large-scale sequencing of diverse breeds will likely expand the catalog of actionable variants, facilitating wider clinical adoption of bioinformatics-driven pharmacogenomics in veterinary medicine.

3.4 One Health Genomics Integration

The One Health framework underscores how animal, human and environmental data converge in disease surveillance and diagnostics (Scarpa and Casu 2024; Pathak and Kim 2024). Bioinformatic integration across species accelerates detection of zoonotic threats. For example, genomic surveillance of bacterial pathogens in livestock – using whole-genome sequencing and resistance gene databases – can preempt antibiotic resistance trends relevant to both veterinary and human medicine (Scarpa and Casu 2024). Similarly, multi-species metagenomic studies track the spread of viruses (e.g. avian influenza sequences in poultry and wild birds) to inform public health risk (Scarpa and Casu 2024). In practice, clinical veterinary sequencing data (from farm outbreaks or pet clinics) are increasingly shared in public repositories, enabling cross-domain analyses of pathogen evolution and host response.

Bioinformatics tools facilitate these One Health linkages. Comparative genomic databases and pipelines can align sequences from human and animal isolates to identify shared variants. For instance, aligning *Mycobacterium tuberculosis* genomes from humans and cattle using phylogenetics has traced zoonotic transmission events (Scarpa and Casu 2024). In precision oncology, the fact that dogs share human environments and have intact immune systems means that canine tumor sequencing data can inform human drug development; bioinformatic meta-analyses combine human and dog cohorts to validate cancer biomarkers (Cahill *et al.,* 2024). Ethical and practical challenges remain (e.g. data privacy, differing disease definitions), but early studies demonstrate the power of transdisciplinary genomics.

In summary, a One Health approach extends veterinary bioinformatics beyond individual animals. It promotes standardized data integration (using common formats and ontologies), shared bioinformatics pipelines and joint interpretation of genomic findings. Through such integrated genomics, bioinformatics in veterinary diagnostics contributes to global health by linking animal cases to broader epidemiological patterns (Scarpa and Casu 2024; Cahill *et al.,* 2024).

3. FUTURE DIRECTIONS

Despite substantial advances, several gaps limit the full potential of bioinformatics in veterinary diagnostics. First, reference genomic resources for many species (e.g. large animals, exotic pets) are still incomplete. The canine reference genome (CanFam3.1) is robust but lags behind human GRCh38 in annotation depth and many livestock breeds lack high-quality assemblies (Pontius *et al.,* 2007; Pathak and Kim 2024). Continued efforts to improve and annotate animal genomes will enhance alignment accuracy and variant calling. Second, large curated databases of normal genetic variation in animal populations are sparse. In humans, databases like gnomAD aid in distinguishing benign polymorphisms from disease-causing mutations; analogous catalogs for dogs, cats and farm animals are emerging but not yet comprehensive (Pathak and Kim 2024; Cahill *et al.,* 2024). Expanded sequencing of healthy and affected animals across breeds will fill this gap.

Third, bioinformatic pipelines validated for veterinary diagnostics are limited compared to human clinical genomics. Many tools (aligners, variant callers, taxonomic classifiers) were developed for human data and their performance on non-human sequences can vary (Resende *et al.,* 2019; Cahill *et al.,* 2024). Rigorous benchmarking of pipelines on veterinary datasets is needed. Along these lines, the use of simulated "spiked" samples or inter-laboratory comparisons (e.g. consensus proficiency tests) can assess reliability in veterinary contexts (Pathak and Kim 2024). Fourth, interpretation of sequence results in animals often relies on human knowledge. While comparative genomics helps, species-specific variant annotation (e.g. linking a mutation to an animal disease) is still an emerging field (Cahill *et al.,* 2024). Development of veterinary-specific variant interpretation guidelines and knowledgebases (akin to ClinVar or CIViC in human oncology) would standardize reporting of diagnostic genomic findings.

Finally, the ultimate clinical impact of veterinary bioinformatics remains under-studied. Few prospective trials have quantified how genomic diagnostics change outcomes in animals. The existing evidence (e.g. Wu *et al.,* 2023; Chon *et al.,* 2023) suggests benefit, but larger multi-center studies are needed. Moreover, cost-effectiveness analyses of genomic diagnostics in veterinary practice are lacking (Scarpa and Casu 2024). From a

bioinformatics perspective, advances in artificial intelligence could enhance variant interpretation and pathogen detection, but require curated veterinary training data.

Future directions include integrating multi-omics (genome, transcriptome, proteome) from animal patients to achieve deeper phenotyping, powered by network analysis and machine learning (Pathak and Kim 2024). The maturation of portable sequencing (e.g. Oxford Nanopore) will bring field-deployable genomics to veterinary diagnostics. Importantly, fostering collaborations between veterinary and human health bioinformaticians will accelerate progress, leveraging the One Health paradigm to share tools and knowledge.

4. CONCLUSIONS

Bioinformatics is rapidly transforming veterinary clinical diagnosis, enabling data-driven insights that were previously confined to human medicine. We have reviewed how sequencing-based diagnostics, supported by robust computational pipelines, are being applied to animal cancer, infectious disease and pharmacogenomics. Comparative oncology research has shown that many genomic findings in pets have direct translational relevance (Wu *et al.,* 2023; Cahill *et al.,* 2024) and specialized cancer panels for dogs meet high technical standards (Chon *et al.,* 2023). Genomic approaches to pathogen detection provide powerful, culture-free diagnosis of zoonoses in animals (Resende *et al.,* 2019; Scarpa and Casu 2024). Early pharmacogenomic successes (e.g. MDR1 genotyping) illustrate the path toward precision medicine in veterinary care (Mealey 2004; Pathak and Kim 2024). All these advances occur within a One Health framework: animal genomic data not only improve veterinary patient management but also inform human health risks (Scarpa and Casu 2024; Cahill *et al.,* 2024).

However, challenges remain in expanding reference data, validating veterinary-specific pipelines and integrating complex multi-omics data. Addressing these gaps will require coordinated research efforts and infrastructure development. Our systematic review highlights the importance of continued investment in veterinary bioinformatics to realize its full potential for animal and public health. By rigorously applying and advancing bioinformatic methodologies, veterinary science will increasingly benefit from the era of precision diagnostics, ultimately improving outcomes for animals and contributing to the health of ecosystems worldwide.

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