Inventorying the 'Precision Medicine' Toolbox

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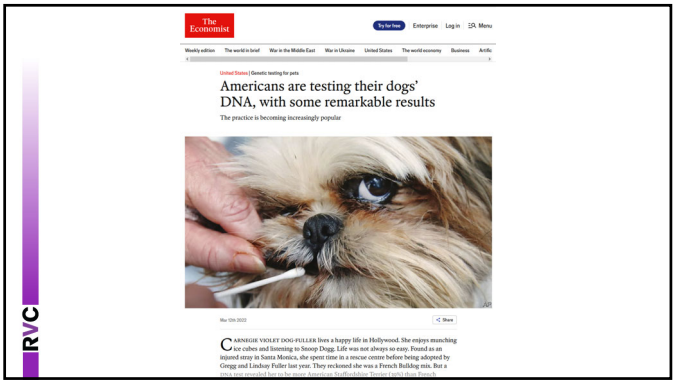
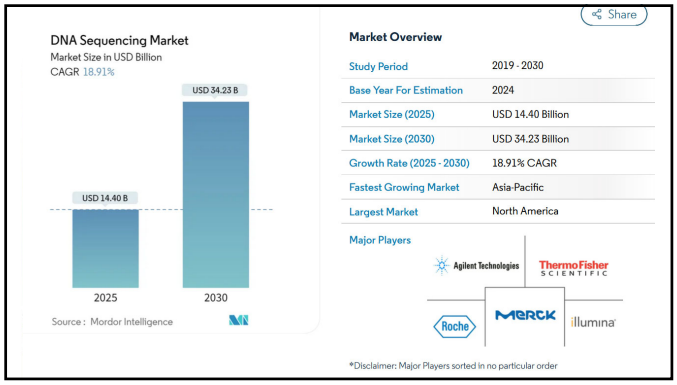
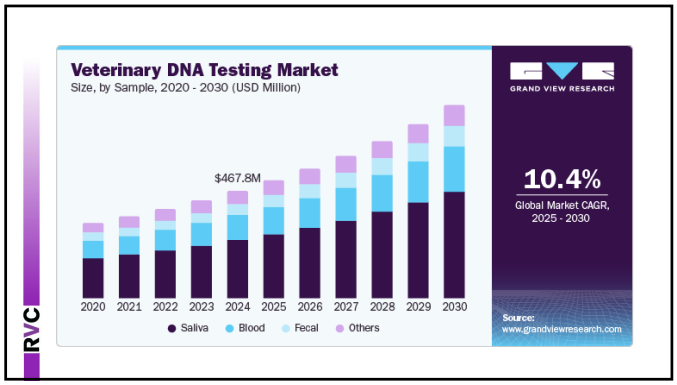
2025 Conti Symposium on Veterinary Continuing Education
Current Concepts and Controversies in Clinical Genomics




DNA Sequencing Market
Market Size in USD Billion
CAGR 18.91%
USD 34.23 B
USD 14.40 B
2025
2030
Source : Mordor Intelligence
Share
Market Overview
Study Period 2019 - 2030
Base Year For Estimation 2024
Market Size (2025) USD 14.40 Billion
Market Size (2030) USD 34.23 Billion
Growth Rate (2025 - 2030) 18.91% CAGR
Fastest Growing Market Asia-Pacific
Largest Market North America
Major Players
Agilent Technologies
ThermoFisher SCIENTIFIC
Roche
MERCK
illumina
*Disclaimer: Major Players sorted in no particular order


Veterinary DNA Testing Market
Size, by Sample, 2020 - 2030 (USD Million)
$467.8M
2020 2021 2022 2023 2024 2025 2026 2027 2028 2029 2030
Saliva Blood Fecal Others
GRAND VIEW RESEARCH
10.4%
Global Market CAGR, 2025 - 2030
Source: www.grandviewresearch.com
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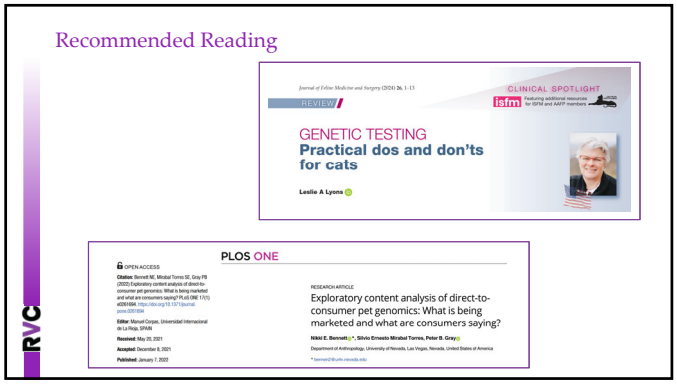
Recommended Reading
REVIEW
GENETIC TESTING
Practical dos and don'ts for cats
Leslie A Lyons
CLINICAL SPOTLIGHT
PLOS ONE
OPEN ACCESS
RESEARCH ARTICLE
Exploratory content analysis of direct-to-consumer pet genomics: What is being marketed and what are consumers saying?
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Overview

- Precision medicine – definition
- What is needed for a precision medicine approach?
- How does genetic background influence disease risk or outcome?
- What commercial tests are available?
- What is transcriptomics?
- What is single-cell biology?
- Summary

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According to the NIH Precision Medicine Initiative, **precision medicine** is :

"*an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person*"

https://ghr.nlm.nih.gov/primer/precisionmedicine/definition

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According to the NHS, 'Personalised medicine' is:

"a move away from a 'one size fits all' approach to the treatment and care of patients with a particular condition, to one which uses new approaches to better manage patients' health and targets therapies to achieve the best outcomes in the management of a patient's disease or predisposition to disease".

https://www.england.nhs.uk/healthcare-science/personalisedmedicine/

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Advantages of a 'precision' approach to veterinary medicine

- ✓ Target interventions only where they are necessary
- ✓ Target interventions only where they are likely to work
- ✓ Avoid 'blanket' therapy in favour of drugs that address pathways more specifically
- ✓ Titrate drugs, doses, durations to the individual patient
- ✓ Identify 'at risk' individuals for screening or close monitoring (early diagnosis)
- ✓ Identify early relapses in susceptible patients
- ✓ Advice to breeders

- > Improved patient welfare
- > Better targeting of healthcare resources (time / money)

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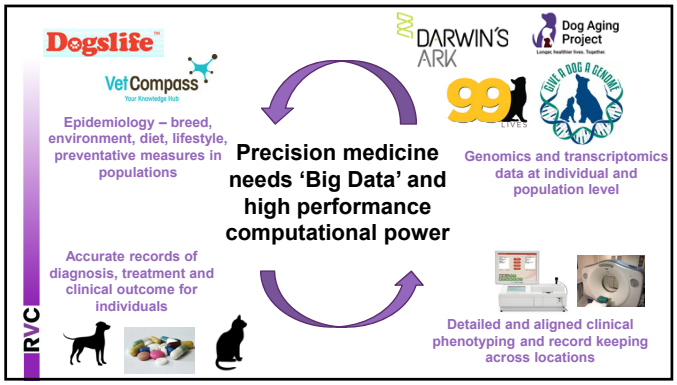

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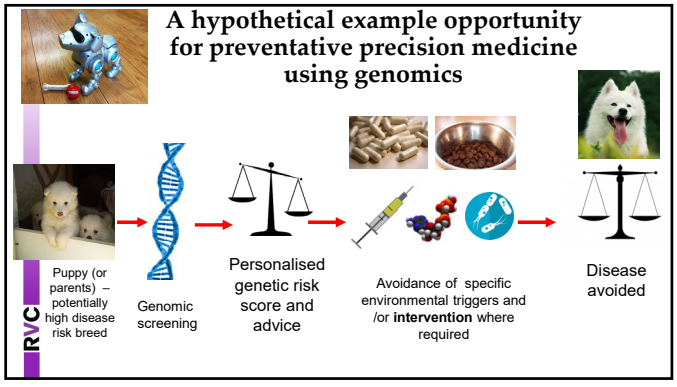

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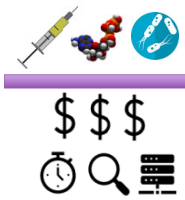
DNA sequence

Without comparative information and expert analysis, this is of limited value



$ $ $

This will look different for each person

Precision Medicine

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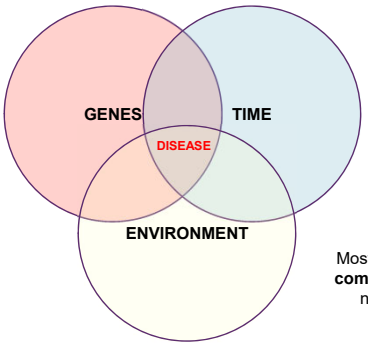

Most diseases have a **complex** origin (ie are not monogenic)

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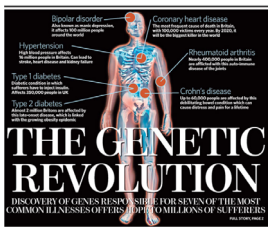
THE INDEPENDENT

Thursday, 7 June 2007



Wellcome Trust Case Control Consortium
***Nature* 2007**

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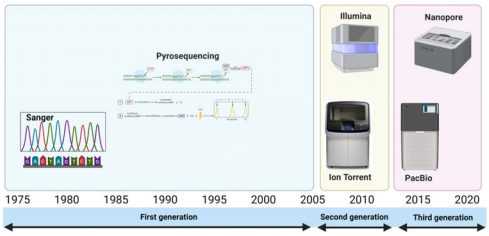
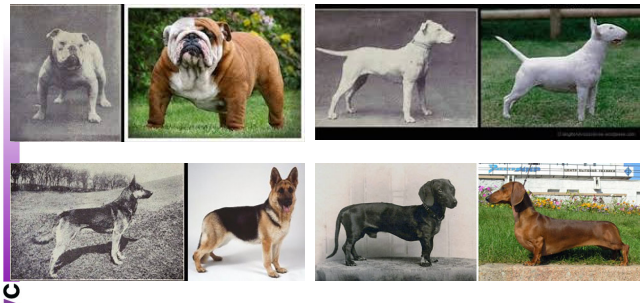
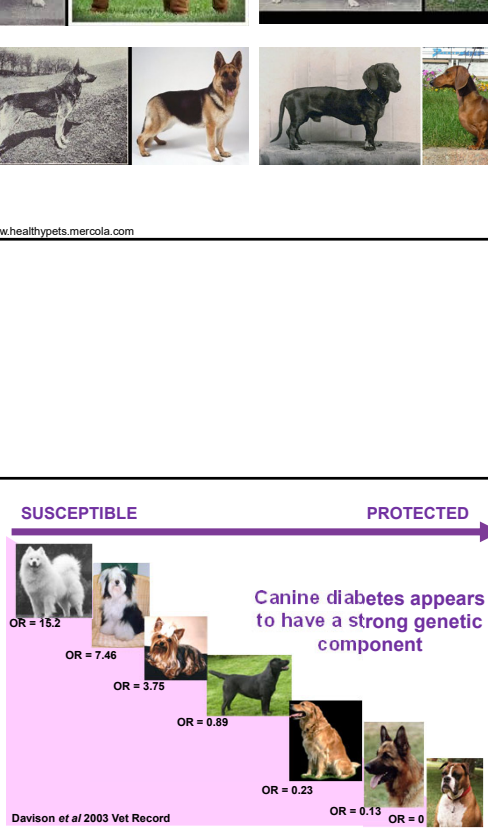
Sequencing technologies

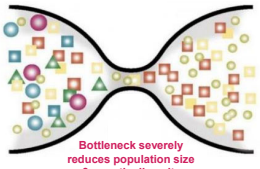


Alsharkski et al 2024
Diagnostics **2024**, *14*(24), 2876; https://doi.org/10.3390/diagnostics14242876



www.healthypets.mercola.com

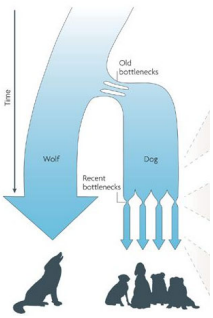




Bottleneck severely reduces population size & genetic diversity

Former Population

New Population



Leader of the pack: gene mapping in dogs and other model organisms

Nature Reviews Genetics 9, 713-725 (2008)

EXAMPLES OF DIRECT-TO-CONSUMER GENETIC TESTS

*Not exhaustive, not intended as a recommendation, not in any particular order

EXAMPLES OF ACADEMIC AND COMMERCIAL VETERINARY GENETIC TESTING FOR CLINICIANS

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Examples of companies and academic institutions offering "genetic testing" for dogs and cats in 2025*:

Table 1. Direct-to-consumer nonhuman animal genetic testing services identified using general internet search query. Using the company's descriptive language, details include the tests offered, species the test is used for, and what the test was advertised to include.

Company Name	Tests Offered	Species	Test Includes
Basepaws	Breed + Health DNA Test	Feline	Breed groups, health, traits, and habits.
Canine HealthCheck	Canine HealthCheck Kit	Canine	Health, phenotype/genotype
DNA My Dog	DNA My Dog NEXTGEN Breed Identification and Genetic Age Test	Canine	Breeds, personality traits, genetic health concerns, predisposition to disease
EasyDNA	Premium Dog Testing Package	Canine	Dog allergy test, breed test, genetic age test
EasyDNA	Cat Genetics DNA Test	Feline	Breed similarities, wild cat index percentage
Embark	Breed + Health Kit	Canine	Health conditions, physical traits, breeds, family tree, relatives
Optimal Selection	Optimal Selection Canine	Canine	Health, Traits Test, Breed, Genetic Diversity
Optimal Selection	Optimal Selection Feline	Feline	Health, Traits
Orivet Genetic Pet Care	Genopet 3.0 (breed + Health Kit)	Canine	Breeds, Health, Lifeplan
Orivet Genetic Pet Care	Cat DNA Health Screen	Feline	Health, LifePlan
Wisdom Panel	Wisdom Panel Premium	Canine	Genetic conditions, breeds, ancestry, physical traits, veterinarian consult

https://doi.org/10.1371/journal.pone.0261694.t001

- https://embarkvet.com/
- https://www.wisdompanel.com/
- https://www.gensoldx.com
- https://www.orivet.com/
- https://basepaws.com/
- https://petdna.ancestry.com/
- https://mycatdna.com/
- https://animalgenetics.com/dog-tests/
- https://vgl.ucdavis.edu/
- https://cvm.missouri.edu/research/canine-genetics-laboratory/canine-genetics-laboratory-testing/

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Why study "Genomics" - how do genes confer risk ?

- Risk is conferred by the inheritance of a particular allele (version) of a gene
- The inherited pair of alleles (e.g. AG, GG or AA) is known as the individual's genotype at that locus
- There are **'monogenic'** diseases in which a mutation in the coding sequence of one gene is responsible for a particular phenotype
- There are also '**complex**' diseases and traits, where multiple genetic risk factors combine with environmental triggers to give disease
- **Understanding which genes contribute to risk can help to identify new treatment or preventative targets**
- **We are a long way behind human medicine in relating DNA sequence to diseases and treatments in veterinary medicine**

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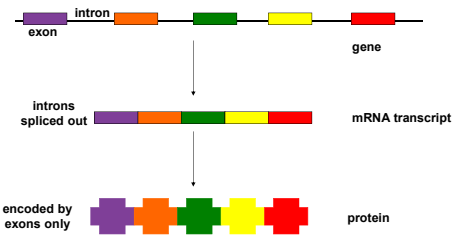
Single nucleotide polymorphisms (SNPs) and disease risk



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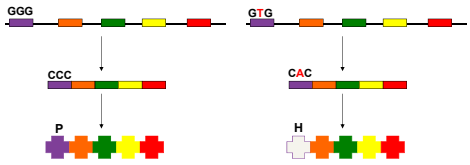
Synonymous and non-synonymous SNPs



A non-synonymous SNP in an exon changes the amino acid (or may insert a STOP codon) in the protein, which can have functional implications

A synonymous SNP does not lead to an amino acid change but may still affect transcript secondary structure, ability to bind a protein or transcript stability

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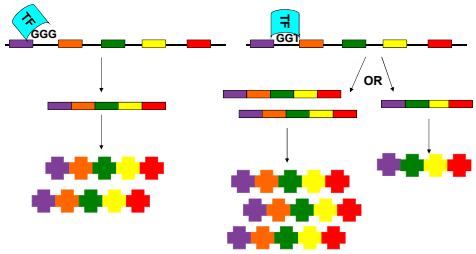
Intronic (non-coding) SNPs can affect transcription factor binding, gene splicing and enhancer activity so may still impact on disease risk



Intronic SNPs may indirectly affect the amount or isoform of a protein produced

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Types of genetic change (other than SNPs) that can affect disease risk or treatment

- Chromosomal
  - Deletions
  - Inversions
  - Duplications
  - Transpositions
- Gene structural variants
  - insertions and deletions (INDELs)
  - copy number variations
  - LINEs and SINEs
- Epigenetic changes

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Types of genetic test on offer

- **ON DNA FROM THE PET THEMSELVES** (genomic DNA) usually from a saliva swab or blood sample e.g.
  - Whole genome sequencing
  - SNP Chip
  - Targeted next generation sequencing for known variants associated with health or appearance
  - PCR tests for specific mutations
- **ON DNA EXTRACTED FROM SOMETHING SAMPLED FROM THE PET** (e.g. tumour, urine, faeces, tonsil swab)
  - Infectious disease – e.g. bacteria, virus, fungus, (PCR)
  - Faecal microbiome
  - Tumour (acquired) mutation analysis

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EXAMPLES OF DIRECT-TO-CONSUMER TEST THAT USE TARGETED GENETIC SEQUENCING

SOME TESTS LOOK SIMILAR BUT ARE NOT GENETIC – these are similar to lateral flow tests for protein, not DNA or RNA

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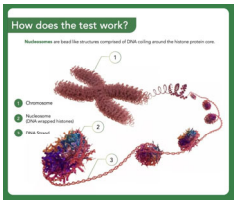
Vidium Searchlight DNA

IDEXX RealPCR panels to detect GI or respiratory infections

EXAMPLES OF GENETIC TESTS DESIGNED TO BE USED WITH VETERINARY INTERPRETATION

Nu.Q® Vet Cancer Test



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So what questions could a "genetic test" answer for a client?

- **General interest:**
  - What breed(s) is my dog or cat?
- **Pre-breeding, pre-purchase or post-purchase health predications in healthy pets**:
  - Does my pet have any inherited diseases for which the gene mutation is known?
  - Is my pet at increased risk of any specific conditions that I can avoid by manging their lifestyle?
  - Is my pet a silent carrier of any mutations that could cause disease in their offspring?
  - Is my pet a silent carrier of any organisms (virus, bacteria etc) that could cause health issues?
  - Does my pet have a diverse and healthy gut microbiome?
- **Diagnostic and treatment questions during illness:**
  - Is my pet's disease caused by a known mutation and can this inform their treatment?
  - Is my pet's disease caused by a particular virus, fungus, protozoa etc that needs specific treatment?
  - Are there genetic mutations in my pet's tumour that inform diagnosis, prognosis or treatment?

WHAT IS IN THE 'GENOMICS' TOOLKIT BEHIND THESE TESTS?

Technology	How is it performed?	Typical sample	Cost	Usual time taken	Need advance knowledge of the mutation, species genome and condition?	Difficult to design or interpret?	Typical uses
PCR (polymerase chain reaction)	Amplify and 'Sanger' sequence a small (known) DNA fragment. Sometimes "multiplex" – looking at several genes at once	DNA from a mouth swab or blood sample	$	Quick	Yes – exact sequence and gene / mutation of interest	+	- Monogenic disease testing (eg PRA) - Infectious disease testing (eg Lepto)

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Targeted sequencing	Use either MassArray technology or NGS and probes to amplify and sequence selected smaller regions	DNA from a mouth swab or blood sample Fecal sample	$$	Days to weeks	Yes – reference sequences around the mutation of interest are required	++	- More commonly a research tool - Can be useful for microbiome analysis (16s rRNA gene)

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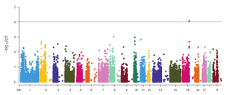
Long-read whole genome sequencing	Oxford Nanopore and PacBio are the two main companies offering this technology. Sequencing reads are in the thousands of base pairs. *MinION version of this technology is designed for 'quick' in the field sequencing		$$$$$	*Weeks to months	Yes – a reference genome and high performance computer required	++++	- Variations in viruses in "real time during outbreaks - Examining microbes and resistance - Sequencing complex or repetitive regions e.g. for structural variants

Genome-wide association study (GWAS) – using a "SNP Chip"

DNA from Cases
n=20 to several thousand

DNA from Controls
n=20 to several thousand

Manhattan plot shows significant SNPs differing in frequency between the two populations



MinION device (Oxford Nanopore) – "pocket sized" genome sequencing technology

Dogslife

VetCompass
Your Knowledge Hub

Epidemiology – breed, environment, diet, lifestyle, preventative measures in populations

Accurate records of diagnosis, treatment and clinical outcome for individuals

DARWIN'S ARK

Dog Aging Project
Longer, healthier lives. Together.

99 LIVES

GIVE A DOG A GENOME

Precision medicine needs 'Big Data' and high performance computational power

Genomics and transcriptomics data at individual and population level

Detailed and aligned clinical phenotyping and record keeping across locations

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"Whole genome sequencing" (WGS):
How much does it cost and how quick is it?

- The first human genome took **$2.7 billion** and almost 15 years to complete.
- The first canine genome was published in 2005 and cost millions of dollars

- **Whole genome sequencing (WGS)** can now be performed on as little as 200ul of whole blood for c.$500 per sample

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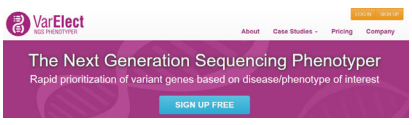
Caveats to WGS for clinical use (in 2025!)

The process takes time, high performance computing facility, an analysis pipeline, and a skilled bioinformatician:

- 'Call' the variants – there will be millions of them!
- 'Annotate' the variants - what predicted impact do they have?
- 'Filter' the variants - to find which are associated with your disease phenotype, and which could play a plausible role in risk using:
  - Other genomes (same breed, relatives)
  - Other databases (gene expression, function, regulation)



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Where and how are we using WGS in veterinary medicine?

- **Individual cases of interesting phenotypes** with many possible candidate genes e.g. clotting disorders, haematological disorders, congenital renal disease
- Potentially **monogenic** traits (e.g. canine juvenile diabetes in Labrador retrievers)
- Differential susceptibility to **infections** e.g. severe coronavirus infections
- **Breed-associated diseases** e.g. Samoyed diabetes, Miniature Schnauzer hyperlipidaemia, Burmese cat diabetes, Greyhound meningitis, CKCS mitral valve disease
- Wide use in **production animal medicine** too – especially cows and poultry, to improve health, welfare and yields

Canine Diabetes GENETICS PARTNERSHIP

MASCOT
Mapping Animal Susceptibility to Coronaviruses: Outcomes and Transcriptomics

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"Transcriptomics" – what can it teach us?

- **This tells us which genes are being expressed in a diseased tissue (compared to healthy tissues)**
- The most common method for transcriptomics analysis is called "RNA-Seq"
- Only a small amount of tissue is needed (e.g. Trucut biopsy)
- Also requires bioinformatics analysis
- Results can guide therapy and identify if particular treatment targets are being expressed
- Can also be compared to survival data to identify why some patients do better than others
- **"Single cell transcriptomics" take things one step further**

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What is RNA-Sequencing?

- ✓ Offers a snapshot of gene expression in the sample, also known as the transcriptome
- ✓ Demonstrates presence and quantity of RNA molecules in the biological sample
- ✓ Uses high-throughput sequencing of RNA extracted from a tissue
- ✓ Relatively unbiased (you do not need to know which genes you are looking for in advance)
- ✓ Requires bioinformatic analysis
- ✓ It is also possible to detect viral RNA and acquired mutations that are present in cancer tissue but absent from healthy tissue

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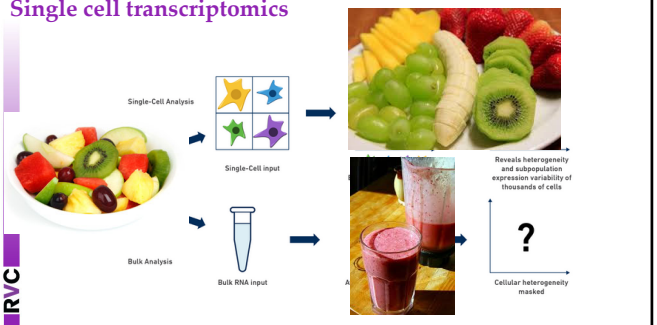
Single cell transcriptomics



https://community.10xgenomics.com/t5/10x-Blog/Single-Cell-RNA-Seq-An-Introductory-Overview-and-Tools-for/ba-p/5

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SCIENCE TRANSLATIONAL MEDICINE | FOCUS

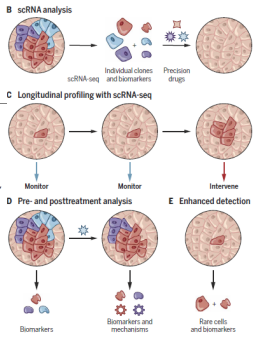
CANCER

Single-cell analyses to tailor treatments

Alex K. Shalek[1,2,3]* and Mikael Benson[4]*

Single-cell RNA-seq could play a key role in personalized medicine by facilitating characterization of cells, pathways, and genes associated with human diseases such as cancer.

One technique of "LIQUID BIOPSY" involves searching for individual cancer cells in the blood stream using sc-RNA-Seq.



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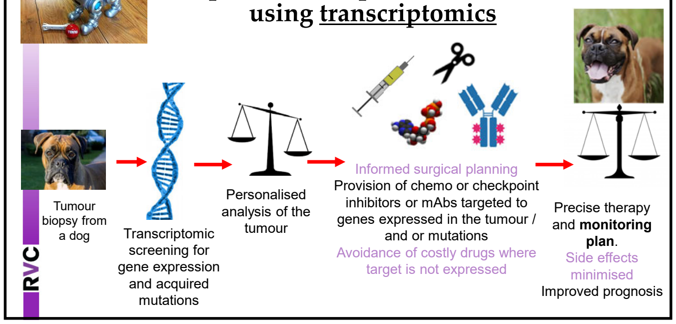
A hypothetical example opportunity for preventative precision medicine using <u>transcriptomics</u>



Overview

- Precision medicine – definition
- What is needed for a precision medicine approach?
- How does genetic background influence disease risk or outcome?
- What commercial tests are available?
- What is transcriptomics?
- What is single-cell biology?
- Summary

Inventorying the 'Precision Medicine' Toolbox

THANK YOU FOR YOUR ATTENTION – QUESTIONS?

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