SASTA OLIPHANT SCIENCE AWARDS

SOUTH AUSTRALIAN SCIENCE TEACHERS ASSOCIATION

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Science Writing

Year 9-10

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DNA: Nature's Secret Code

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Year 10 2025
10-14 High Street Kensington, Mary MacKillop College
Word count: 1360

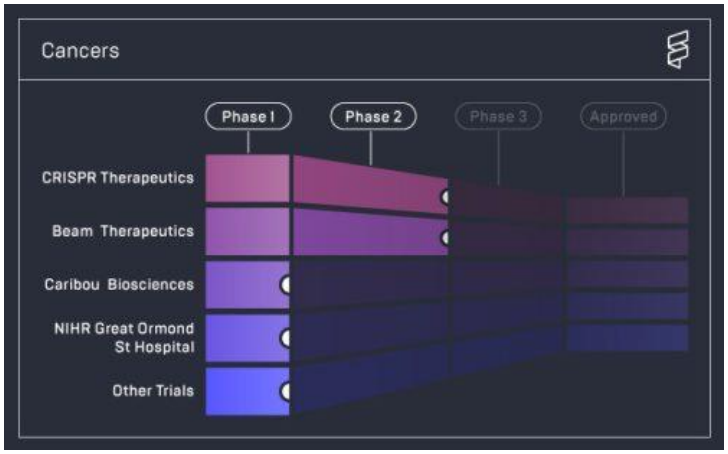
Unlocking the secret code of DNA.

Beginning with a humble pea plant experiment, Gregor Mendel, often referred to as 'the father of genetics', paved the way for the discovery of DNA, which has only in the past 10 years been found to have groundbreaking applications into modern medicine. As medicine advances, DNA increasingly correlates to pharmacogenomics, cancer research and treatment, vaccine development, prenatal care, recombinant DNA technology, disease prevention, past DNA analysis, and DNA targeted pharmaceuticals. These are merely a few examples for what DNA in a medical setting could behold, with the American National Cancer Institute stating that "there are tremendous potential applications for CRISPR-Cas [Clustered Regularly Interspaced Short Palindromic Repeats] and their derivative systems (i.e. dCas9) due to the ability to accurately determine the underlying disease causes, genetic mutation variants, immunological regulatory factors, cell signalling mediators, and drug targets as well as drug molecules and therapeutics" (National Library of Medicine, 2021) furthermore, "there's also hope that [CRISPR] will have a place in treating cancer" (National Health Institute, 2020). Understanding the past and present of DNA will enable us to unlock the extraordinary potential of DNA in medicine, as in the words of Stanford researcher Stanley Qi, "I am confident that CRISPR will become a pillar of medicine." (Stanford, 2024).

"In 1902, Sir Archibald Edward Garrod became the first person to associate Mendel's theories with a human disease." (DNA worldwide, 2015). He later published his findings in recessive inheritance. His work is now considered the cornerstone of genetic history. In the words of the American National Center for Biotechnology, Sir Garrod "can rightly be deemed one of the most profound intellectuals of the 20th century, whose bequests to science and medicine continue to increase in value." (National Center for Biotechnology Information, 2008). As a result of these discoveries, in 1959, a scientific breakthrough was made in the connection between an additional copy of chromosome 21 and Down's syndrome. In the late 1960's-1970's, cytogenetics became more widely accredited, including the Giemsa stain; named after German bacteriologist and chemist Gustav Giemsa for the stain used in diagnosing malaria and other parasites (National Library of Medicine, 2007). In 1983 was when Huntington's disease – a progressive, neurodegenerative disease, became the first mapped genetic disease, with a genetic marker found on chromosome four, although the gene was not fully isolated until 1993 (DNA Worldwide, 2015). In 1990, the first gene associated with increased risk of breast and ovarian cancer was identified as BRCA 1 located on chromosome 17. With further study, it became clear that a second gene BRCA 2 located on chromosome 13 was also involved. The reason being that BRCA 1 and BRCA 2 were both identified as tumour suppressor genes, with scientists concluding that an altered copy of either gene may lead to the accumulation of mutations, leading to cancer (National Cancer Institute, 2024). To capitalise upon major advancements made in genetic medicine, The National Human Genome Research Institue launched The Human Genome Project. This was an endeavour to map the human blueprint comprising of international scientists, which successfully revealed the first sequence of the human "genome sequence that accounted for over 90% of the human genome" (National Human Genome Research Institute, 2024). In March 2022, the first truly complete human genome was announced in by the Telomere-to-Telomere consortium. Since then, the concepts of using DNA in medicine have gained traction, especially since genomics played a factor in the covid vaccine, and are now being involved in tailoring pharmaceuticals, anaesthesia, treating cancer and genetic disorders. The success of genetic medicine today has only been made possible due to the efforts of the numerous past studies conducted.

President Bill Clinton and Francis Collins, M.D., Ph.D., at the White House celebrating the draft human genome sequence generated by the Human Genome Project. Dr. Collins served as the de facto leader of the International Human Genome Sequencing Consortium, the group that sequenced the human genome during the Human Genome Project. (Human Genome Project, 2022)

In 2025, DNA is being taken into serious consideration in healthcare internationally. In Australia, "the Genomics Health Futures Mission is investing $500.1 million in genomic medicine research over 10 years" (Australian Government, 2025). More than ever now, gene altering technology such as CRISPR, and SeekRNA are making headway. CRISPR is a Nobel Prize winning discovery that may be likened to a pair of molecular scissors. CRISPR has the incredible ability to direct Cas-9 enzymes, a macromolecule drug, to cut through genes, allowing qualified personnel "to fix a mutated gene or regulate a defective gene to treat a disease" (Stanford, 2024). On "November 16, 2023, the UK's Medicines and Healthcare Products Regulatory Agency approved Casgevy for the treatment of SCD and TDT." (Henderson H, 2024). This was followed by the United States FDA approval for the genetic editing in treatment of Sickle Cell anaemia, and beta thalassemia. In Australia, The University of Sydney has labelled SeekRNA "a pathway for accurate gene editing" - "beyond CRISPR" (University of Sydney, 2024), in terms of accuracy utilising a programmable ribonucleic acid strand - whereas CRISPR relies on "other proteins or the DNA repair machinery" (University of Syndey, 2024) - that can identify locations for insertion in gene sequences, therefore vastly reducing the error factor. Both of these cutting-edge technologies play a major role in precision medicine - the application of gene editing in genomic medicine, as a result of CRISPR, SeekRNA and further research, "genomics has exploded from widespread sequencing availability to TGA-approved therapies requiring a precise genetic diagnosis in less than a decade" (The Medical Journal of Australia, 2022), bringing a surge of demand in the genomics medical field, and the updating of medical training curricula to include genomic treatment.



2024 visual representation of CRISPR in clinical trials to be approved for public usage, Henderson, H 2024, *CRISPR clinical trials: a 2024 update*, Innovative Genomics Institute (IGI), viewed 15 March 2025, <https://innovativegenomics.org/news/crispr-clinical-trials-2024/>.

The future of genomics in medicine is taking shape with National plans globally, one of which being Australia's National Framework for Genomics in Cancer Control, which was released in February of this year. Similarly, National strategies have also been enacted in the United States, United Kingdom, Germany, Canada, France, Japan, Switzerland and Finland, among others. With scientific and medical advancements of this magnitude, there have been numerous ethical and equitability concerns. One major act in Australia surrounding genetic editing is the Prohibition of Human Cloning for Reproduction Act 2002, in which "section 15 of the PHRC Act specifically prohibits a person from altering the genome of a human embryo in such a way that the alteration is heritable by descendants *and* the person intended this to be so" (University of Melbourne, 2018). However, there are still ethical concerns, for instance the long-term effects of gene editing, safety - as there may be unintended errors akin to CRISPR cutting a gene off target causing cancerous cells, mosaicism; the occurrence of some cells being edited, while others remain original, financial concerns such as the cost of genetic medical treatments mean that they are widely unavailable to the middle class and below. Other ethical concerns raised are; confidentiality, discrimination - if genetic information is utilised in such way as to determine suitability for insurance, or employment and the autonomy of future generations (National Human Genome Institute, 2017). Regarding these concerns, there are regulations such as the PHRC, nonetheless, as the science advances, so does the law. Through vigorous selection, certain treatments are being made publicly available, "supported by state-based funding, and further prompted strategic implementation projects; for example, the NSW Health Genomics Strategy, which facilitated the first TGA-approved clinical *in vivo* gene therapy in Australia for retinal dystrophy and gene therapy for spinal muscular atrophy in newborns." (The Medical Journal of Australia, 2022). Reflecting the growth of genomics in medicine, and as a whole endeavour, "the global genomics market size was valued at USD 33.90 billion in 2023 and is poised to grow from USD 39.53 billion in 2024 to USD 157.47 billion by 2033" (BioSpace, 2024)

Ultimately, the study of DNA has triumphed every expectation, leading humanity to be within inches of unlocking its secret code. Starting as Mendel's humble pea experiment, DNA research has blossomed into studies that encompass the treatment for thousands of diseases and impairments, and in some cases, may even cure them. As we advance into the future, the applications of genomics into medicine are revolutionary, with life-changing therapeutics CRISPR and SeekRNA poised for clinical implementation on the international scale. Despite lingering ethical and equity concerns, the progression of genomics is undeniable. The future of medicine and healthcare is now deeply integrated with genomic studies, and as we begin to decode the mystery of DNA, life itself, the potential for curing human disease, impairments, and conditions is finally within our grasp.

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