JULY | 2021

BIO-BABBLE

Newsletter of the Australasian Biospecimen Network Association

ABNA UPDATES

- A reminder that ABNA memberships are due for renewal on July 31st. Invoices have been sent out to current members for renewal.
  - All financial ABNA members qualify for reduced registration fees for the annual conference and are eligible to vote during the ABNA Annual General Meeting.
  - Pricing details are available on the ABNA website **HERE**

- Are you advertising to fill a biobanking position? Are you holding a biobanking event our members can attend? If you would like ABNA to circulate your job ad or event details get in touch with us. We aim to not overrun your inbox and will limit these mailouts to once a week - so please get in touch as early as possible on info@abna.org.au

- Your ABNA Conference Organising Committee is currently putting together the draft schedule for the Hybrid event.
  - The conference website will be live in August
  - Call for abstracts will open in September

- Let the ABNA newsletter sub-committee know if there is an aspect of biobanking you would like to see featured in Bio-Babble or if you would like to provide an article for our readers. A reminder that this newsletter is sent out on the last Thursday of the month with the content deadline set a week prior.

#ABNA2021Virtual

BIOBANKING GREATEST HITS - A RETROSPECTIVE
PART ONE

By Cassandra Griffin

1: BRACA in Black

In 1974 Mary-Clare King began her hunt for the genetic underpinnings of breast cancer following earlier revelations that women with a first-degree relative with breast or ovarian cancer were at a higher risk of disease themselves. Given PCR would not be invented for another 9 years and genome was not yet a recognised term this was a huge undertaking. The work began using a large cohort of epidemiological data collected from 1579 breast cancer patients and used complex segregation analysis to demonstrate that familial clustering could be explained by an autosomal dominant trait. In order to allow for the development of linkage maps - based on the frequencies of recombination between known genetic markers during the crossover of homologous chromosomes - Dr King and her colleagues recruited 329 relatives with 146 cases of invasive breast cancers and banked relevant blood samples from each individual. In 1990 after extensive analysis she presented the results at the American Society of Human Genetics Meeting and in 1991 the gene was dubbed BRCA1.

Even though Dr. King and her colleagues did not win the race to clone BRCA1, her work in compiling this original collection and uncovering evidence of a genetic cause completely shifted the view of the role of genes in cancer.

Album: Back in Black
Artist: AC-DC
Release date: 25 July 1980

Album: Who are you
Artist: The Who
Release date: 18 August 1978

2: Who Are You?

The identification of human remains belonging to missing persons is one of the main challenges for forensic genetics. In November 1998, the Spanish Ministry of the Interior decided to support an initiative from the University of Granada to implement a National Program (The Phoenix Program) to attempt to identify cadavers and bones from missing persons.

The Phoenix program contains two independent databases (the questioned database, containing STR profiles and mtDNA sequences from biobanked forensic or autopsy specimens such as bones, and the reference database, containing STR profiles and mtDNA sequences from relatives). The databases house genetic data that can be compared automatically to associate matching or related profiles, such as those from unknown remains and reference samples from relatives.

Participants who consent to the provision of reference samples provide two buccal swabs with a minimum of 2 and a maximum of 4 relatives sampled. All samples are bar coded and banked and subsequent genetic data are coded to maintain confidentiality and reduce the misuse of genetic data. For questioned database samples, typically 2-4 fragments of at least 25 g of compact bone and/or teeth from non-identified cadavers and human remains retrieved from the Guardia Civil biological archives.

To date, >3,700 families have contacted the Phoenix program, 862 have and at least 319 unidentified remains have so far been identified and returned to their relatives. When mtDNA and/or STR associations are found, a second independent analysis is performed as part of the quality assurance process.

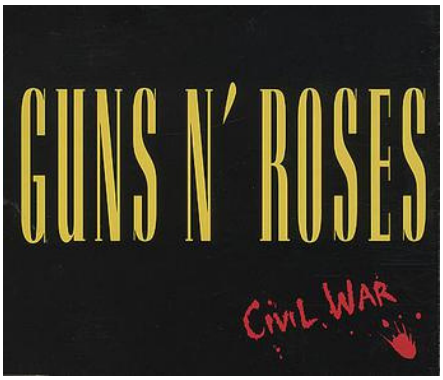
3. Sticky Fingers

Genome-wide association studies, supported by large scale biobanked collections, are used to associate specific gene variants with particular diseases. Scores of genes have been causally implicated in monogenic forms of diabetes but GWASs have now identified over 100 common variant signals.

There have been a number of breakthroughs in Type Two Diabetes (T2D) research which highlight the diverse routes by which human genetic studies, supported by large scale collections, can inform translational medicine. For example the combination of common-variant GWASs and candidate-gene resequencing has demonstrated that loss-of-function mutations in SLC30A8 which encodes a zinc transporter expressed in pancreatic islets are protective for T2D, leading to efforts by several pharma companies to develop ZnT-8 antagonists for therapeutic use.

Furthermore, the combination of -omic measurements, longitudinal clinical phenotypes, and GWAS data has highlighted sets of molecules (e.g., branched-chain amino acids) that not only are prospectively associated with T2D progression but may play a causal role in T2D development, potentially providing a valuable biomarker for early detection.

Album: Sticky Fingers
Artist: Rolling Stones
Release date: 23 April 1971



4. Civil War

In the last 5 years, GWASs have been undertaken for nearly all major immune-mediated diseases (with sample sizes of tens of thousands of biobanked case and control samples for more common immune-mediated diseases. The results obtained through analysis on these vast sample cohorts have made key contributions to deeper biological understanding of immune-mediated diseases

As a flow on effect, analysis of these cohorts have proven highly successful at initiating medication repositioning such as the repositioning of biological medications targeting components of the IL-23 pathway. These are now mainstay treatments for psoriasis and psoriatic arthritis, are noted to be highly effective in ankylosing spondylitis and are effective in IBD.

Many other biobank supported GWAS discoveries have stimulated targeted therapy-development programs including conclusive evidence that immunological reactions to epitopes that had been citrullinated by PAD enzymes were causatively involved in Rheumatoid Arthritis (RA). This led to programs developing PAD inhibitors in RA, and these have shown significant promise.

Single: Civil War
Artist: Guns N' Roses
Release date: 3 May 1993

5: The White Album

Although psychiatric diseases had a slow start in GWAS locus identification, concentrated efforts in psychiatric and neurodegenerative disease biobanking (including recent efforts to recruit for post-mortem brain banking protocols) have resulted in more than 50,000 samples genotyped in the last 5 years and the identification of more than 100 risk loci discovered.

Despite this progress, no new molecular targets for schizophrenia have been successfully identified since the first antipsychotic drugs were identified. This has been attributed to the focus for most drug development remaining on achieving high-potency drugs for a single target. While this methodology has been successful in many other areas of medicine, in schizophrenia it which necessitates a choice between the competing hypotheses of schizophrenia pathophysiology. Recent data have provided unequivocal evidence of polygenicity, and because many of the GWAS loci contain genes that code for proteins among those indicated through multiple prior hypotheses, e.g., dopamine, glutamate, immune modulation, calcium signalling, and nicotinic cholinergic, future drug development may benefit from taking a multi-target approach.

Album: The Beatles
Artist: The Beatles
Release date: 22 November 1968

THE BUSINESS MATRIX – ONLINE FORUM

17th August 2021, Tuesday, 3:30 - 4:30pm (AEST)

Session One Topic - "Professionalism and Career Prospects"
Session Chair - Dr Wayne Ng - Victorian Cancer Biobank

Having dedicated professional staff with the right blend of technical, managerial, regulatory, and ethical skills and experience to meet the challenges of running a sustainable biobanking business cannot be overemphasised. An identifiable career path contributes to the retention of, along with coaching and mentoring of employees in moving forward in this emerging market.

To join us on this free virtual forum, please register **HERE**

CASSANDRA GRIFFIN manages NSW Regional Biospecimen Research Services which includes Hunter Cancer Biobank and the Mark Hughes Foundation Brain Bank at the University of Newcastle, NSW. She has held this position since early 2018 and has contributed to the growth and diversification of the Biobank with support from NSW Health Pathology, the University of Newcastle's Hunter Cancer Research Alliance and Hunter Medical Research Institute. She is currently the vice president of ABNA.

PAUL HOFMAN MD, PhD is professor of pathology and the head of the laboratory of clinical and experimental pathology at Nice Center Hospital, University Côte d'Azur, France. He is the director of the Nice Côte d'Azur Biobank and of the MSc Biobanks and Complex Data Management. He is the director of the OncoAge and of the Research team 4 at the IRCAN Center at the Comprehensive Cancer Center Antoine Lacassagne, Nice. He is a member of the Royal Academy of Medicine, Brussels, Belgium.

Dr ZISIS KOZLAKIDIS is the Head of Laboratory Services and Biobanking at the International Agency for Research on Cancer. He is responsible for one of the largest and most varied international collections of clinical samples in the world, focusing on gene-environment interactions and disease-based collections. This WHO infrastructure supports multinational efforts in making treatments possible and delivering those to resource-restricted settings. Dr. Kozlakidis has significant expertise in the field of biobanking and has served as President of ISBER.

If you have any suggestions for a short article for Bio-Babble, please contact: info@abna.org.au
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