

#### Background In 1948, a man was found mysteriously dead on Somerton Beach, Adelaide, South Australia. His identity remains unknown until this day, and the case has been classified as one of Australia's biggest unsolved

who he actually was. This project analyses Somerton Man's DNA file extracted from his hair which has been corrupted. The project aims to investigate the Somerton Man's DNA with other sample DNA files via computer techniques and biological engineering methods.



Figure 1: The Somerton Man

## **Aim and Motivation**

- To find possibilities of who the Somerton Man was taking a step forward to solving the unsolved mystery
- To evaluate the robustness of the Somerton Man's DNA
- To evaluate the ethinicity of Somerton Man
- To identify any possible diseases or physical characteristics of the Somerton Man

### **Task 1: Counting** DNA

Firstly, the Somerton man's DNA file was examined and the available SNPs to be used for analysis were counted.

There are more than 0.6 million SNPs in Somerton man's DNA file, but only about 2% of them have determined base pairs.

/

# Azizul Hakim Luqman Ul Hakim Ng and Zihe Wang

rs7537756

Supervisor: Prof. Derek Abbott | Co-supervisor: Dr Andrew Allison | EEE | 141 | TT20 | 2019

# THE UNIVERSITY Who killed the Somerton Man?

mysteries. There was no ID or anything on him that shows a clue on



# **Task 2: Ethnicity**

Ethnicity check via GEDmatch shows that he was North Atlantic for a proportion of more than a quarter of the chart. The second largest section shows that he was Baltic, which does not stray too much from North Atlantic region.





#### Figure 5: Change of ethnicity percentage after degrading to certain amount

There is only slight change on the ethnicity regions during the degradation process. It is shown in Figure 5 that the ethnicity does not intersect with one another for two sample DNA files, thus concludes that the degradation of DNĀ does not affect the proportion of ethnicity.

This then concludes that the Somerton Man's origin is around North Atlantic countries and Baltic region based on Figure 4. The countries that are associated with these regions are shown in Figure 6.



Figure 6: North Atlantic and Baltic regions

## **Task3: Genetic Disease**

Somerton Man's DNA was analysed with dbSNP

575 potential genetic diseases were found associated to Somerton Man's DNA.

There is no result strongly support Somerton Man's known physical appearence such as hair colour, teeth structure or eye colour. But several interesting characteristics were discovered.

One of the diseases found in his DNA is Skin fragility woolly hair syndrome which indicates that Somerton Man might have woolly hair abnormality.



Figure 7: Part of potential genetic diseases

## Conclusion

Task 1: The proportion of Somerton Man's DNA is quite low to conduct most DNA analysis services. But there still are some techniques can be tested with it.

Task 2: The Somerton Man might be North Atlantic according to the ethnicity check on GEDmatch

Task 3: No strong evidences to confirm his physical charateristics and genetic diseases. But several interesting results were discovered.

#### R ef er ence

Figure 1: J. Bineth, "How the Somerton Man played cupid from the grave," ABC News. [Online]. Available: https://www.abc.net.au/news/2017-12-14/somerton-man-cold-case-could-be-one-step-closer-tosolved/9245512.

Figure 6: "World Map - Simple," MapChart. [Online]. Available: https://mapchart.net/world.html.

	rs28763969					Curre Released	nt Build July 9,	d 15 , 201	
	Organism	Homo sapiens	lomo sapiens		Reported in <u>ClinVar</u>				
type 1	Position	chr6:7583652 (GRCh38.p12) 🖸		Gene : Consequence DSP : Synonymous Variant					
	Alletes	T>C	2		2 citations				
	Variation Type	SNV Single Nucleotide Va	/ Single Nucleotide Variation		See ra on genome				
	Frequency	C=0.00969 (2435/25130 C=0.00983 (1234/12556 C=0.00962 (1164/12102	14, GnomAD_exome) 18, TOPMED) 18, ExAC) ( <u>+ 8 more</u> )						
nia nemia somal dominant, 3	Variant Details	Alleie: C (alle	Allele: C (allele 10: <u>54105</u> )				0		
	Clinical Signifi	cance ClinVar Accession	* Disease Names			Clinical Significa	nce		
	Frequency	RCV00003807	RCV000038074.6 not specified			Benign		1	
	Allases	RCV00028638	11 Ectodermal dysplasia si	kin fragility syndrome		Likely-Ber	ign		
inant 3		RCV00034109	3.1 Epidermolysis bullosa,	lethal acantholytic		Likely-Ber	ign		
	Submissions	RCV00037848	RCV0X0378485.1 Skin fragility woolly hair syndrome   RCV000384052.1 Arrhythmogenic right ventricular cardiomyopathy.			Likely-Benign Likely-Benign			
arre o	History	RCV00038405							
	Publications	RCV00047731	9.3 Arrhythmogenic right w with woolly hair and ke	Arrhythmogenic right ventricular cardiomyopathy, type 8,Dilated cardiomyopathy with woolly hair and keratoderma			Benign		
ninant, 3		RCV00061730	0.1 Cardiovascular phenoty	/pe		Benign			
/Recessive		RCV00077602	8.1 Cardiomyopathy			Benign			