



THE UNIVERSITY
of ADELAIDE

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Project 141: Who Killed the Somerton Man

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Outline

- **Introduction**
 - **Background**
 - **Project Aims and Motivation**
- **Specific Tasks**
 - **Task 1**
 - **Task 2**
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- **Reference**

Introduction

Background

- Who?

The Somerton Man

- What?

Found dead

- When?

December 1st, 1948

- Where?

Somerton Beach, SA

- How?

Unknown

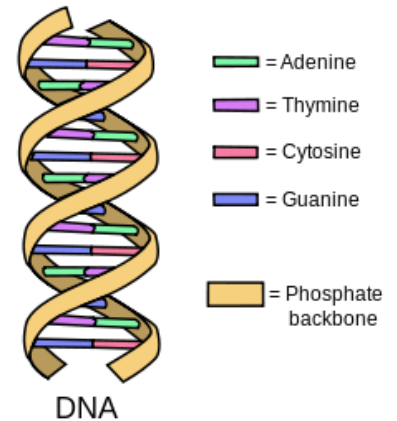


Background (cont.)

Somerton Man's DNA

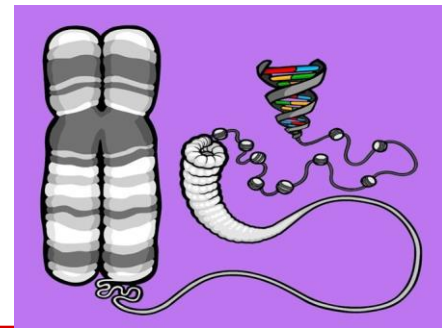
- Hair sample in the Police Museum
- Extract DNA from the hair sample

Background (cont.)



Deoxyribonucleic Acid (DNA)

- Carrying genetic instructions
- A code formed from a chain of 4 chemical bases (nucleotides): adenine (A), guanine (G), cytosine (C) and thymine (T)
- DNA forms chromosomes



Background (cont.)

Single Nucleotide Polymorphism (SNP)

- A variation of base pairs at a specific position in a DNA sequence
- Defines characteristics of individuals (eg. eye colour)

Background (cont.)

Somerton Man's DNA file

#rsid	chromosome	position	genotype
rs548049170	1	69869	--
rs13328684	1	74792	--
rs9283150	1	565508	--
i713426	1	726912	--
rs116587930	1	727841	--
rs3131972	1	752721	--
rs12184325	1	754105	--
rs12567639	1	756268	--
rs114525117	1	759036	--
rs12124819	1	776546	--
rs12127425	1	794332	--
rs79373928	1	801536	--
rs72888853	1	815421	--
rs7538305	1	824398	--
rs28444699	1	830181	--
i713449	1	830731	--
rs116452738	1	834830	--
rs72631887	1	835092	--
rs28678693	1	838665	--
rs4970382	1	840753	--
rs4475691	1	846808	CT
rs72631889	1	851390	--
rs7537756	1	854250	AG
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Project Aims 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 identify any possible diseases and physical characteristics of the Somerton Man
- To find the relatives, and finally find out who he actually was

Specific Tasks

Specific Task

Task 1: Testing with Somerton Man's DNA reference file

Aims

- Counting amounts of SNP
- Try to conduct DNA analysis, and check the results

Specific Task

Task 1: Testing with Somerton Man's DNA reference file

Method

- Writing code to count the SNPs of the DNA files with programming language (eg. C++, JAVA)
- Upload the file to [genesis.gedmatch.com](https://www.genesis.gedmatch.com) (GEDmatch) which provide DNA analysis services



- GEDmatch is a website that has an open data personal genomics database and provide tools for DNA and genealogy research
- Tools used:
 - One-To-Many DNA Comparison
 - One-To-One DNA Comparison
- 2000 SNPs minimum requirements for uploading DNA file

Specific Task

Task 1

Current outcomes

- Counting results of SNPs

Counting results of SNPs			
Chromosome	Total amount	Exist amount	Percentage
1	49510	1014	2.05%
2	51771	978	1.89%
3	43023	658	1.53%
4	39473	621	1.57%
5	37028	661	1.79%
6	44021	880	2.00%
7	34356	655	1.91%
8	31681	601	1.90%
9	26445	519	1.96%
10	30522	705	2.31%
11	30943	705	2.28%
12	29432	596	2.03%
13	22080	393	1.78%
14	19961	441	2.21%
15	19006	440	2.32%
16	20396	558	2.74%
17	19401	519	2.68%
18	17674	372	2.10%
19	14879	514	3.45%
20	14781	375	2.54%
21	8607	245	2.85%
22	8915	303	3.40%
Total	613905	12753	2.08%

Specific Task

Task 2: Artificially complete the Somerton Man's DNA file

Aims

- Create synthetic DNA files based on Somerton Man's file with different strategies
- To see if there are any people in DNA public database linked to the artificial DNA
- To see how close these artificial DNA files are related

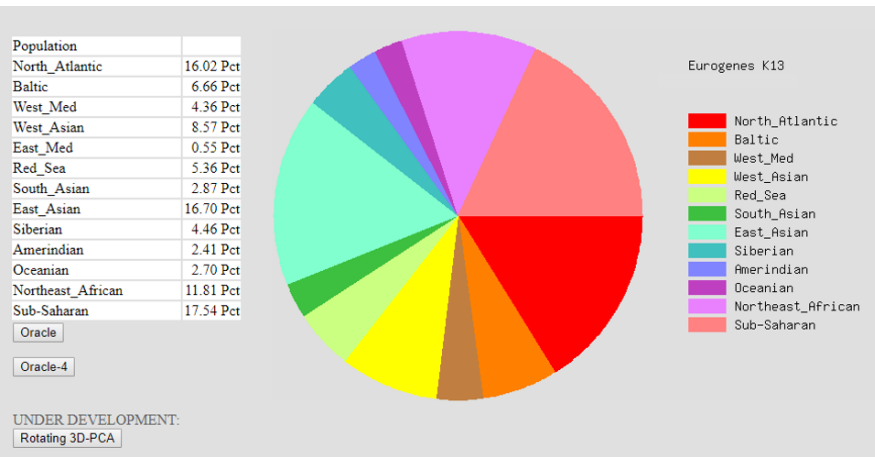
Specific Task

Task 2: Artificially complete the Somerton Man's DNA file

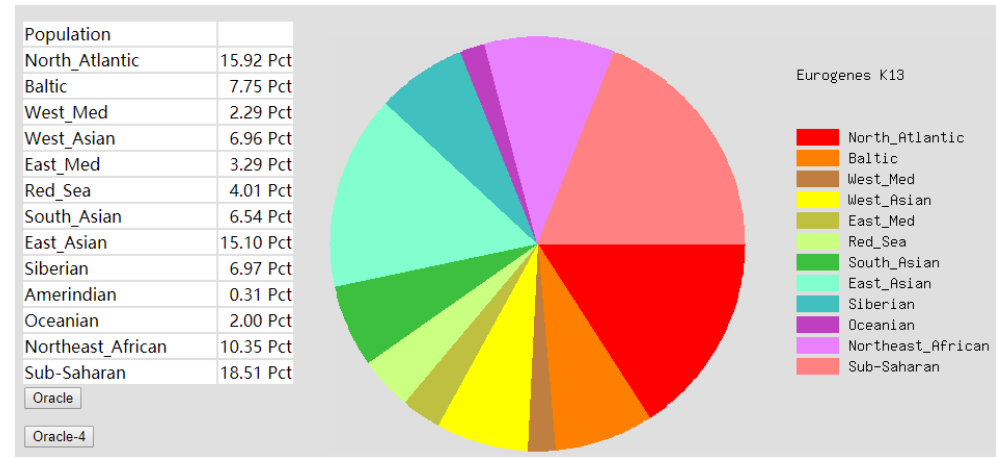
Method

- Writing code to artificially complete the DNA file
- Strategies include replace empty SNPs with random SNPs, homozygous pairs (eg. AA, GG, TT, CC) and etc.

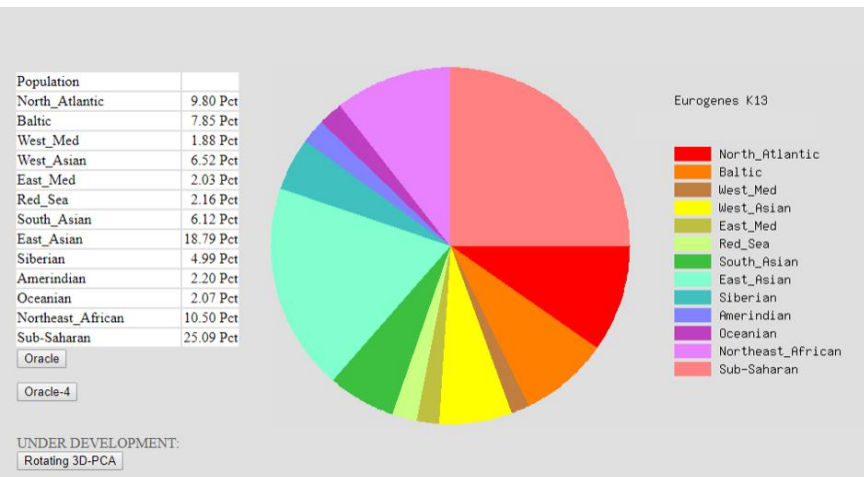
Current Outcome: Ethnicity check



3500 SNPs with uniform pairs for each chromosome



3500 SNPs with random pairs for each chromosome



Tool used: Eurogenes K13
Admixture Proportions

full SNPs with random pairs

Specific Task

Task 2: Artificially complete the Somerton Man's DNA file

Method (cont.)

- Use SNP database and find which SNPs are common to males or have a high likelihood in males. Insert these SNPs into the original files
- Use SNP database and pick some SNPs which have known effects (eg. eye color, hair color). Insert into the original files
- Developing a program to search the information(effects) of each SNPs in the Somerton Man's DNA file

SNP database (dbSNP)

Reference SNP (rs) Report

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rs12913832

Current Build 152
Released October 2, 2018

Organism	<i>Homo sapiens</i>	Clinical Significance	Reported in ClinVar
Position	chr15:28120472 (GRCh38.p12) ?	Gene : Consequence	HERC2 : Intron Variant
Alleles	A>G	Publications	92 citations
Variation Type	SNV Single Nucleotide Variation	Genomic View	See rs on genome
Frequency	G=0.45329 (56919/125568, TOPMED) A=0.4419 (13667/30926, GnomAD) G=0.177 (888/5008, 1000G) (+ 3 more)		

Allele: G (allele ID: [19784](#)) [?](#)

ClinVar Accession	Disease Names	Clinical Significance
RCV000005011.4	Skin/hair/eye pigmentation, variation in, 1	Association

Variant Details

Clinical Significance

Frequency

Aliases

Submissions

History

Publications

Specific Task

Task 3: Test with complete DNA reference files

Aims

- Obtain complete reference DNA files
- Degrade the complete files and test the results from GEDmatch
- Observe the change of results when degrading files to lower level
- Use the complete DNA reference files to modify the Somerton Man's DNA file

Specific Task

Task 3: Test with complete DNA reference files

Method

- Complete DNA reference files in 23andMe format
- Writing code to degrade the complete DNA files to different levels.
- Compare analysis results from GEDmatch and identify differences

Specific Task

Task 3: Test with complete DNA reference files

Method (Cont.)

- Extract SNPs with same base pairs among the complete DNA reference files
- Add these SNPs to the Somerton Man's file and test it on GEDmatch

Project Management

Task allocation

Task	Allocation
Proposal Seminar	Together
Thesis (Draft)	Individual
Project Management	Zihe
Research Method and weekly reports	Azizul
Task 1: Testing with Somerton Man's DNA reference file	Zihe
Task 2: Artificially complete the Somerton Man's DNA file	Together
Task 3: Test with complete DNA reference files	Together
Exhibition Poster	Together
Final Seminar	Together
Project Exhibition	Together
Honours Thesis/Final Report	Individual
YouTube Video	Azizul

Risk Assessment

Risk	Likelihood	Consequences	Risk Estimation
Absence of meeting	Unlikely	Minor	Low
Miscommunication of members	Unlikely	Moderate	Medium
Loss of data	Unlikely	Severe	High
Delay of task completion	Likely	Major	High
Bugs in codes	Likely	Minor	Medium
Out of budget	Rare	Severe	Medium
Misunderstanding of tasks	Unlikely	Moderate	Medium
Unethical works	Unlikely	Major	Medium
Member drop the course	Rare	Severe	Medium
Bad quality of purchased items	Unlikely	Major	Medium

Milestones

Semester 1	Milestone	Semester 2	Milestone
week 5	Complete Task 1	week 1	Review works
week 6	Proposal Seminar Slides	week 9	Complete Task 3
break	Proposal Seminar	week 12	Exhibition of projects
Week 11	Complete Task 2	Week 12	Final Thesis
Week 12	Thesis Draft	Week 13	Final seminar

Reference

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Thank you



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