UNIVERSITÀ
DEGLI STUDI DI TRIESTE

dipartimento
di ingegneria
e architettura

Corso di Laurea in Ingegneria Clinica e Biomedica
Informatica Medica I

IL CASO DI STUDIO

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What is Nutrigenomics

- Diet has a key role in influencing the risk of chronic diseases
- The genetic background can alter the host's physiological response to diet

Nutrigenomics is the application of high throughput genomics tools in nutrition research based on

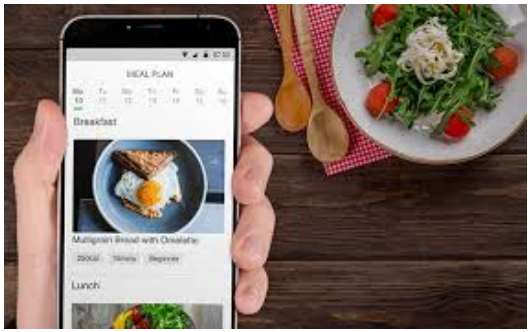
a **systematic collection** and analysis of **nutritional data and their correlation to genotypes, phenotypes and lifestyle**

Nutrigenomic research

→NUTRIGENOMIC RESEARCH aims to provide personalized dietary/lifestyle guidelines:

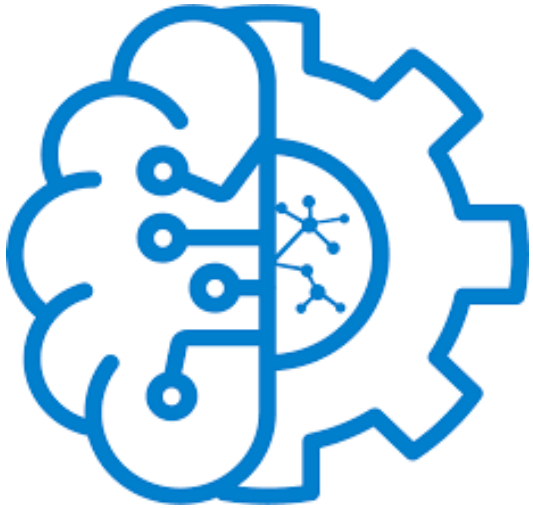
- To protect public health
- To reduce modifiable risk factors
- To indicate choice of food

Supporting nutrigenomic research: data collection and outputs



Input

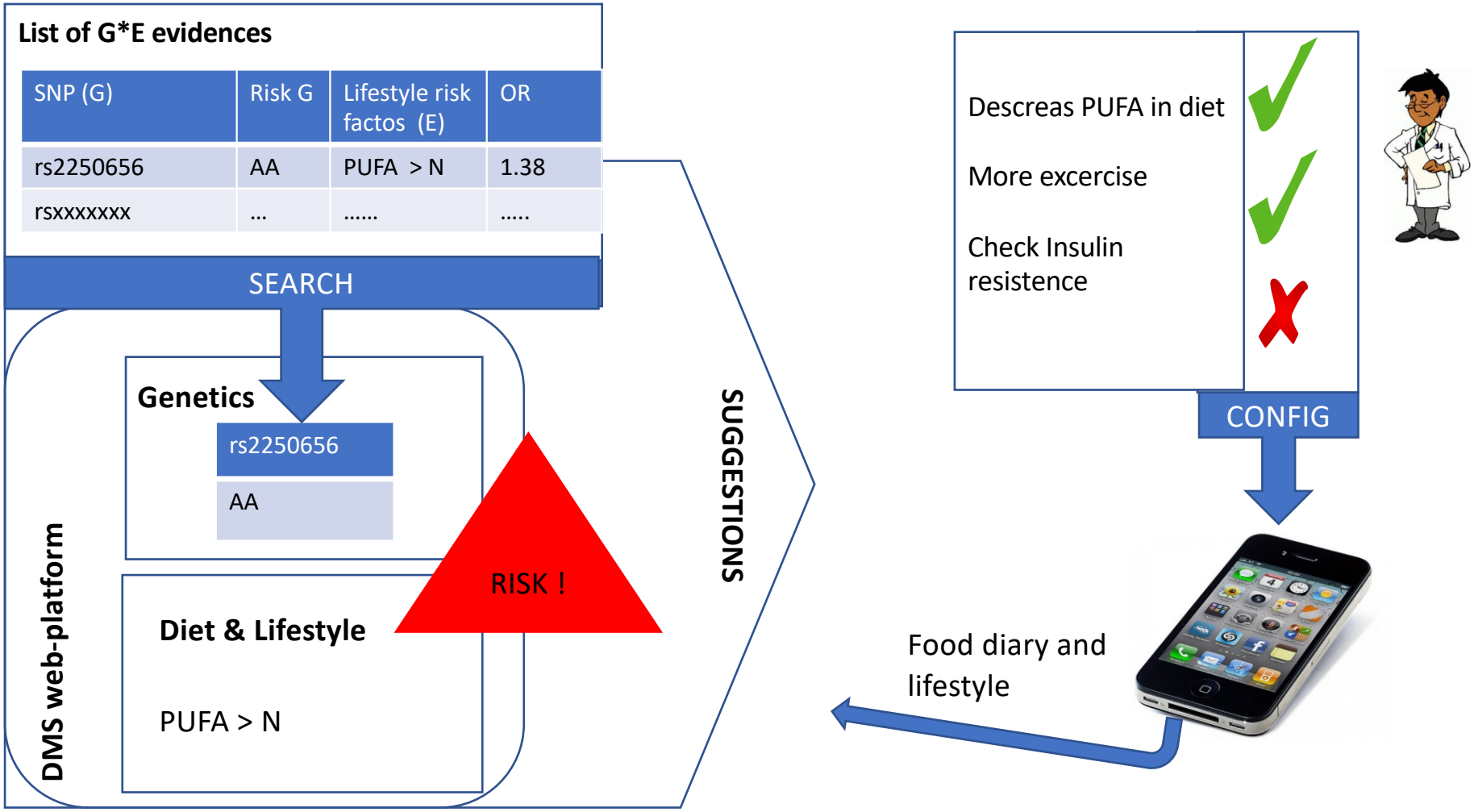
Lifestyle of individuals
Biomarkers
Current diet
Phenotype data
Genotype data



Output

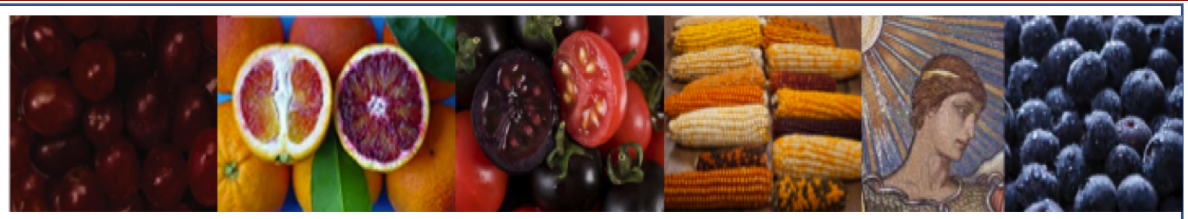
Personalized optimum lifestyle plan
- Menu plans
- Exercise regimens
- Medical recommendations
- Personal follow-up medical plans

Decision Support Systems



Case study: the ATHENA project

ATHENA project
funded by European Commission, 7FP, 14 Partners



AnThocyanin and polyphenols bioactive for Health Enhancement through Nutritional Achievement

Epidemiological study to determine interaction between anthocyanin consumption, genetic structure and cardiovascular risk

Thourough clinical assessment (N=500)	Dietary assessment 24h recall 4 times: one per season	Genotyping

Side-project: Collaboration in the validation of a questionnaire to assess adherence to mediterranean diet

DIETARY ASSESSMENT TOOLS: the Food Atlas to select quantities (1/2)

"Today I ate two bowls of dog food, a sandwich crust, some spaghetti that fell on the floor, half of your cat food, a wet tea bag, three bugs and the inside of a sneaker. How many grams of fat is that?"

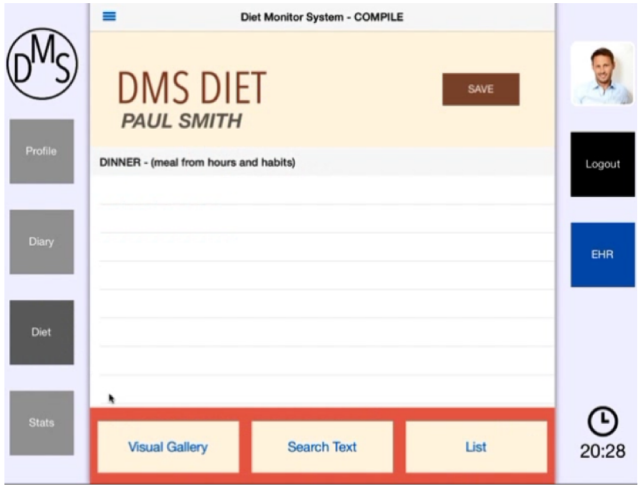
DIETARY ASSESSMENT TOOLS: the Food Atlas to select quantities (1/2)

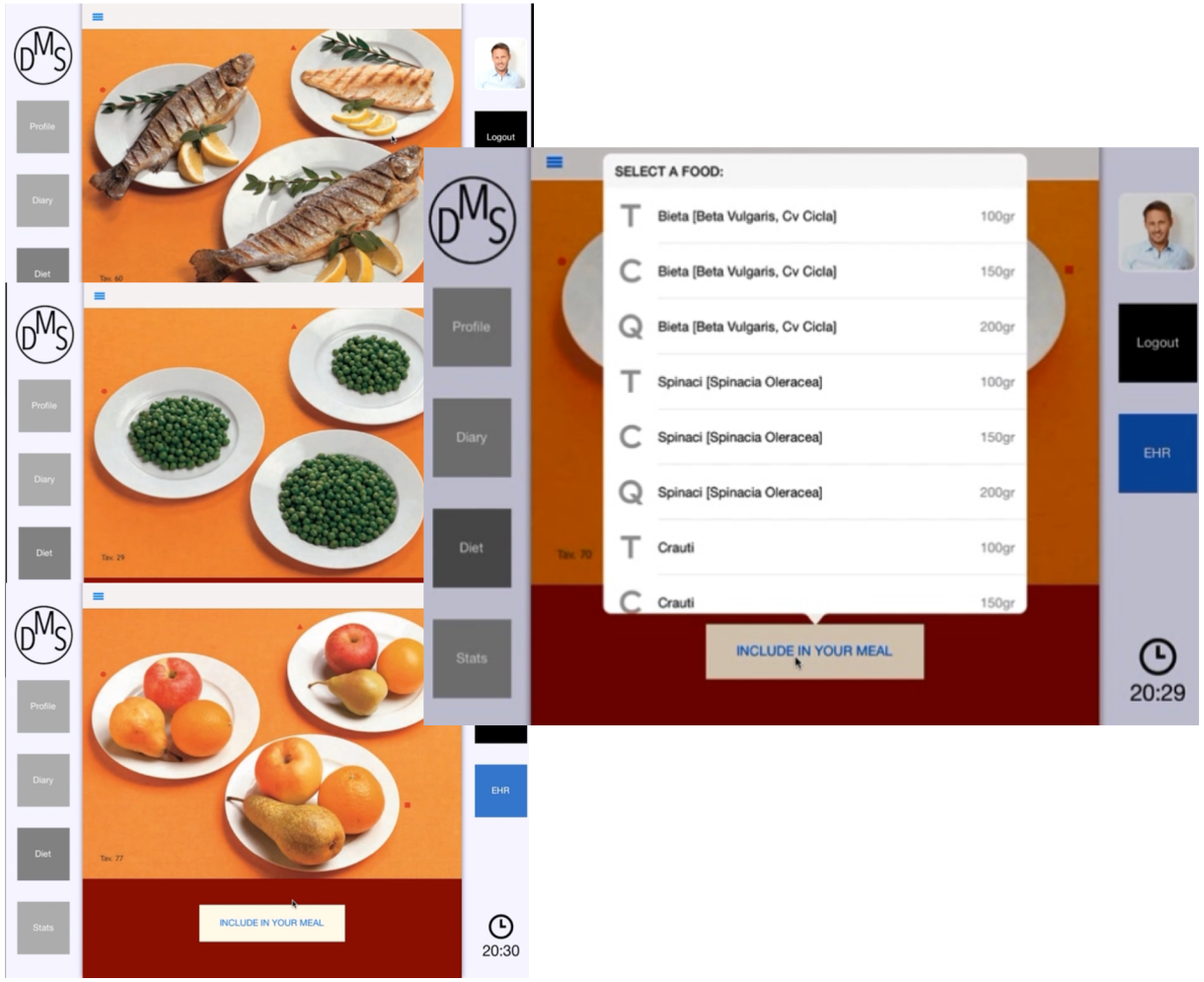
What the interviewed subject sees:

What the dietician sees:

		grams
▲	Mixed salad	50
●	Mixed salad	100
■	Mixed salad	150

The mHealth App for the patient at home: food diary





Evaluation of micro- and macro-nutrients

- ADDA (Athena diet data analysis) allows calculating the composition in micro and macro nutrients of the diet.
- It combines input data collected during dietary interview and the micro and macro nutrient composition for each food from food databases.

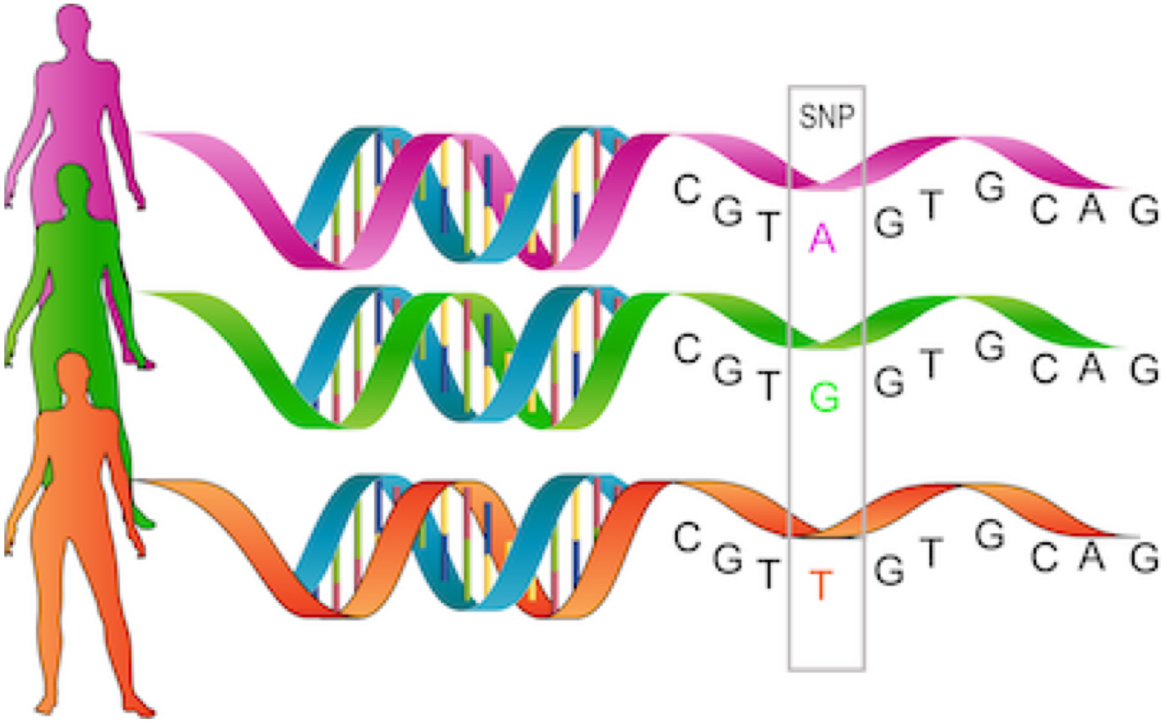
ID	Date	TIME	g Water	g Proteins	g Vegetable proteins	g Total lipids	mg Vitamine C	mg Anthocyanins
GAL-11	15/05/2013	Breakfast	510,45	6,01	2,29	3	1,35	0
GAL-11	15/05/2013	Snack	22,61	19,95	0	19,6	0	0
GAL-11	15/05/2013	Lunch	259,68	15,48	6,6	28,18	81,5	0
GAL-11	15/05/2013	Snack	2,73	0,84	0,84	0	0	0
GAL-11	15/05/2013	Dinner	1914,13	43,24	8,34	39,91	14	9,05
GAL-11	15/05/2013	TOTAL	2709,6	85,52	18,07	90,69	96,85	9,05

Food Composition Database to calculate micro and macro nutrients

Food Composition Database for Epidemiological Studies in Italy (IEO) enriched with: anthocyanins contents of additional foods and food items used by vegans/vegetarians

Food Code	Food Name	edible part	Energy, recalculated	Total protein	Vegetable protein	Total fat	Animal fat	Vitamin B1, Thiamin	Vitamin B2, Riboflavin
		g	kcal	g	g	g	g	mg	mg
381	POTATOES	83	85	2,1	2,1	1	0	0,1	0,04
50399	BATATAS or SWEETPOTATOES	84	87	1,2	1,2	0,3	0	0,17	-2
380	POTATOES, YOUNG or EARLY	96	67	2	2	0	0	0,12	0,03
3002	STARCH, POTATO	100	349	1,4	1,4	0	0	-2	-2
100219	POTATO, POWDER	100	318	9,1	9,1	0,8	0	0,04	0,14
382	POTATO CRIPS, PLAIN	100	531	7	7	34,6	0	0,17	0,2
18	TAPIOCA	100	363	0,6	0,6	0,2	0	0	0,1
303	ASPARAGUS, WILD FROM WOOD	57	35	4,6	4,6	0,2	0	0,13	0,43
304	ASPARAGUS, WILD FROM FIELD	87	29	3,6	3,6	0,2	0	0,21	0,29
305	ASPARAGUS, GREENHOUSE	52	24	3	3	0,1	0	0,27	0,25
700484	ASPARAGUS, canned	100	18	2,1	2,1	0,7	0	0,06	0,1
8035	ALFA ALFA SPROUTS	100	24	4	4	0,7	0	0,04	0,06
350	SOYA, SPROUTS	98	49	6,2	6,2	1,4	0	0,23	0,2
306	BEETROOT	82	19	1,1	1,1	0	0	0,03	0,02
312	CARROTS	95	33	1,1	1,1	0	0	0,04	0,04
8032	DAIKON	87	15	0,8	0,8	0,1	0	0,03	0,02
341	TURNIP	69	18	1	1	0	0	0,02	0,07

GENOTYPING DATA: SNPs

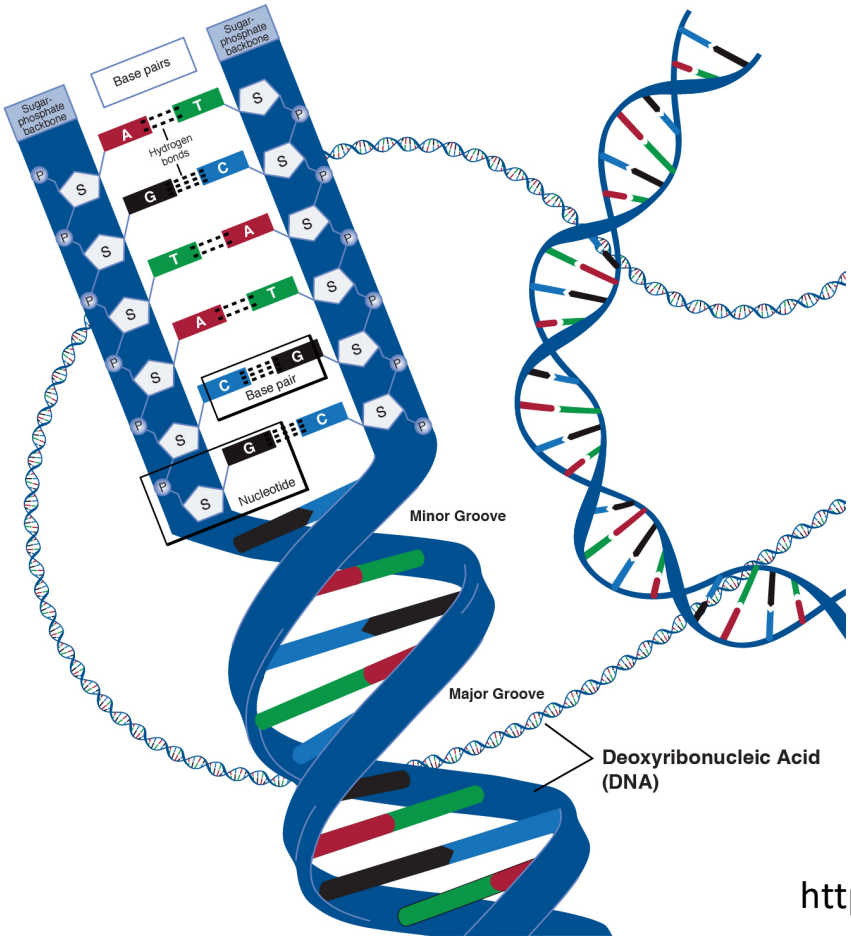


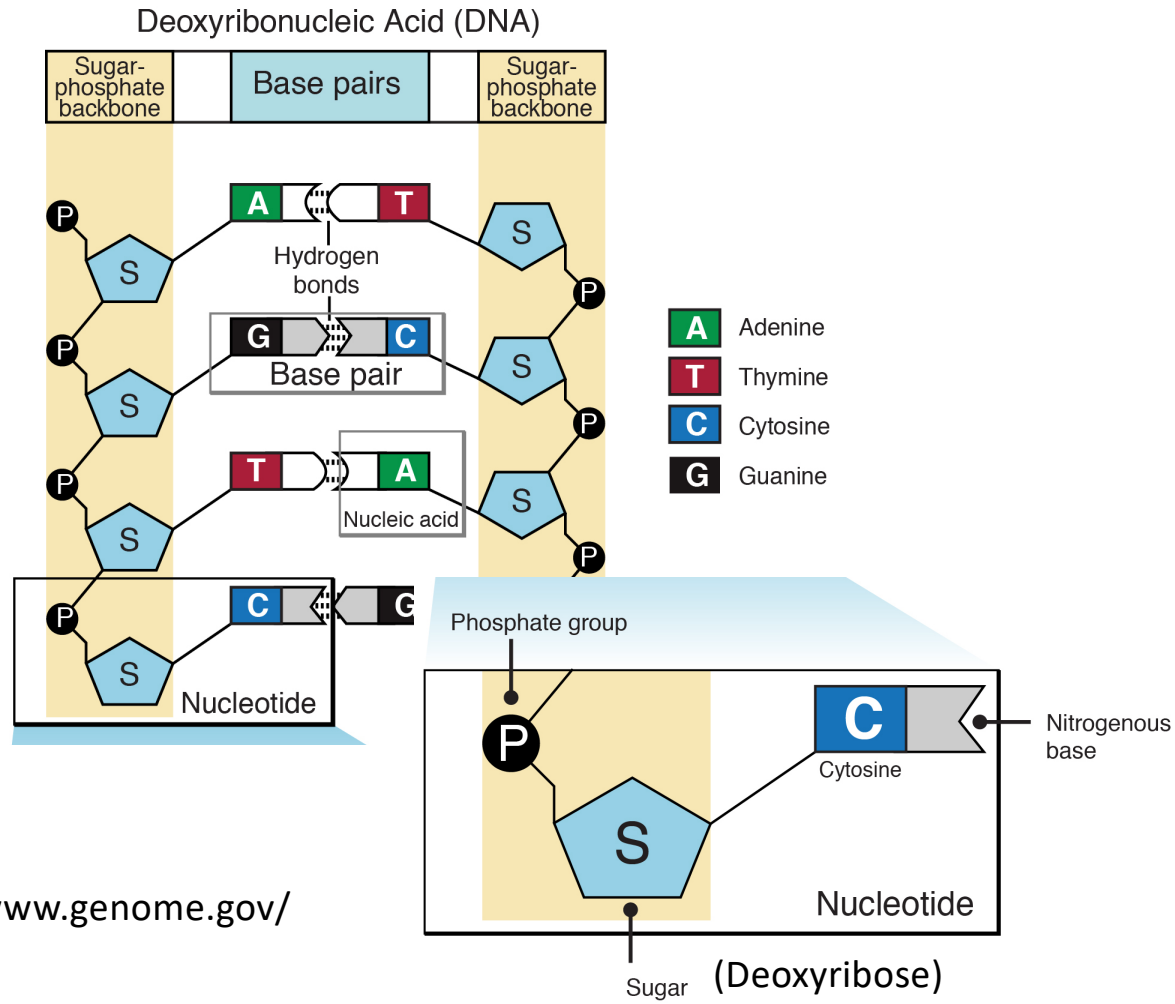
Single Nucleotide Polymorphism

Change of a nucleotide in a specific position of the genome

It has a frequency depending on how often we find a specific variant in the population

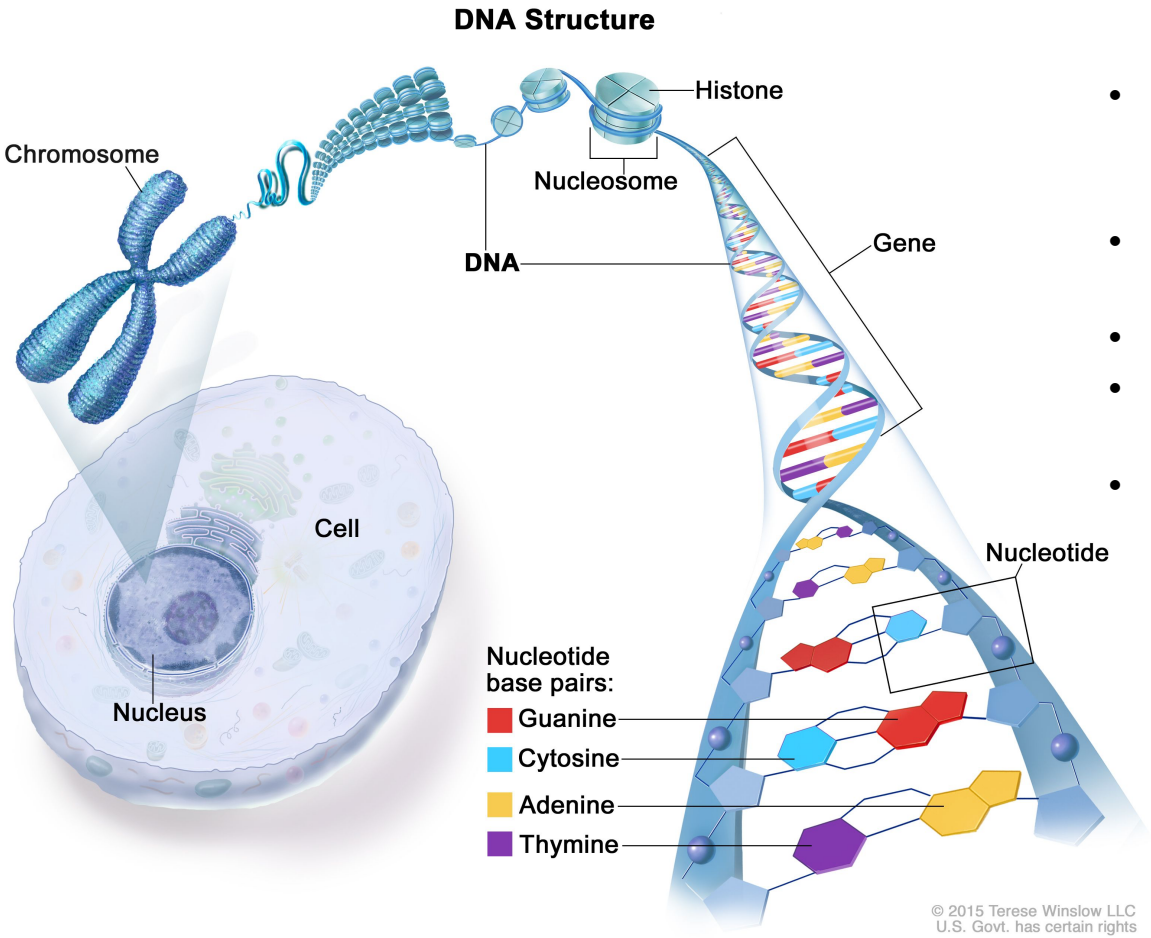
The DNA structure





https://www.genome.gov/

The DNA structure



- **Chromosomes** are paired (we have two copies of the same gene in two different chromosomes)
- Humans have 23 pairs of chromosomes--22 pairs of numbered chromosomes, called autosomes, and one pair of sex chromosomes, X and Y.
- Each parent contributes one chromosome to each pair
- An **allele** is one of two or more versions of a gene.
- An individual inherits two alleles for each gene, one from each parent.
- If the two alleles are the same, the individual is homozygous for that gene. If the alleles are different, the individual is heterozygous.

https://www.genome.gov/

Representation of SNPs

Individual 1

Chr 2 ...CGATATTCC**T**ATCGAATGTC...
copy1 ...GCTATAAGG**A**TAGCTTACAG...

Chr 2 ...CGATATTCC**C**ATCGAATGTC...
copy2 ...GCTATAAGG**G**TAGCTTACAG...

SNP heterozygous (AG)

Individual 2

Chr 2 ...CGATATTCC**C**ATCGAATGTC...
copy1 ...GCTATAAGG**G**TAGCTTACAG...

Chr 2 ...CGATATTCC**C**ATCGAATGTC...
copy2 ...GCTATAAGG**G**TAGCTTACAG...

SNP homozygous with two major (GG)

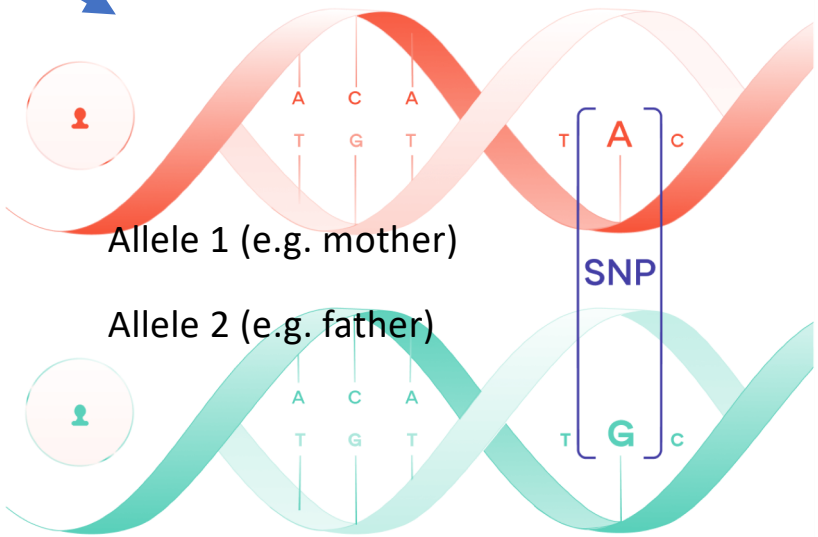
Individual 3

Chr 2 ...CGATATTCC**T**ATCGAATGTC...
copy1 ...GCTATAAGG**A**TAGCTTACAG...

Chr 2 ...CGATATTCC**T**ATCGAATGTC...
copy2 ...GCTATAAGG**A**TAGCTTACAG...

SNP homozygous with two minor (AA)

Minor allele = base with the lowest frequency



Allele 1 (e.g. mother)

Allele 2 (e.g. father)

Major allele = base with the highest frequency

GENOTYPING DATA: ped and map files

Genotyping data: ped and map files (e.g. Illumina Infinium HumanCore)

- map: SNPs heading

Chromosome | Position in centimorgans | Base position

Chr	SNP	cM	pb
1	rs12565286	0	711153
1	rs28659788	0	713170
1	rs11804171	0	713682
1	rs2977670	0	713754
1	rs12138618	0	740098
1	rs3094315	0	742429
1	rs3131972	0	742584
1	rs3131968	0	744055
1	rs1048488	0	750775
1	rs12562034	0	758311
1	rs2905035	0	765522
1	rs12124819	0	766409
1	rs2980319	0	766985

- ped: Inviduals genetica data

The rows of the .map file represent the columns of the .ped file

Standard heading | SNPs described in map file

family ID	individual ID	father	mother	sex	affection status	Genotypes				
GAL_11	GAL_11	0	0	2	0	G G	C C	T T	C C	G G

Allele 1 Allele 2

OUR MATERIAL

- .xls table with clinical data
- map and ped files (reduced version)
- Paper with a previous analysis of the data (Rizzi F et al, 2016)
- Reference papers on nutrigenomics