

NuGO  
Wageningen, Netherlands  
Sept 2011

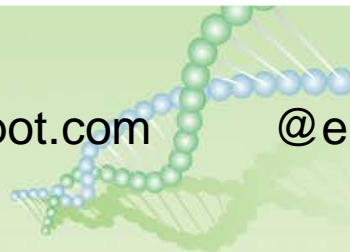


**“Personal Genetics in Practice”**

Keith Grimaldi Ph.D  
ICCS, NTUA, Athens

[Eurogene.blogspot.com](http://Eurogene.blogspot.com)

@eurogene



# Wayback...

INTERNET ARCHIVE  
**WayBackMachine** BETA

4 captures  
13 Nov 01 - 27 Jun 02

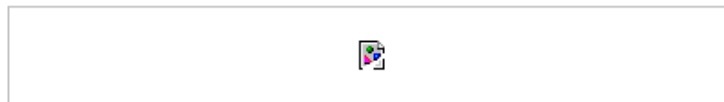
OCT NOV DEC  
2000 **13** 2001 2002

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Beyond the Edge



[hot off the press](#) » [news archive](#)

## Consumer Gene Testing Service is Coming From UK's Sciona Ltd

**August 15 2001**  
BioWorld International  
by Nuala Moran

LONDON - Sciona Ltd. says it will be the first company to offer a consumer gene testing service, screening for single nucleotide polymorphisms affecting metabolism and providing personalized nutrition and dietary

Sciona has had discussions with the UK government advisory body, the Human Genetics Advisory Commission, and the Medical Device Agency about the launch of the service, and the need to create a regulatory framework for such testing services. "We want to be regulated, as we believe this will help to improve public confidence in our service," Martin said.







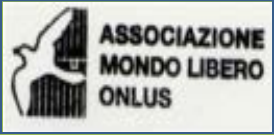







# Administrative Information



- **Project Acronym:** EUROGENE
- **Project Title:** An integrated high secure cross-border platform for the delivery of optimal personalized lifestyle recommendations based on genetic analysis
- **Contract:** eTen Market Validation: 046310
- **Budget:** € 1,730,335
- **Starting date:** 1<sup>st</sup> January 2008
- **Ending date:** ~~30<sup>th</sup> June 2009~~ December 2009



# The consortium...

Partner	Country	Role in the Project	Short Name
		Administrative Coordinator Customisations - Technology	ICCS
		Scientific and Pilot Coordinator -Service Provider NB: SCIONA is not active in the project since Feb 2009	Sciona
		Pilot – Focus: CVD (~75 patients)	Mondo
		Business & Market Opportunities	INI
		Pilot – Focus: Obesity (~100 patients, 50 obese vs. 50 lean)	Bonn
		Pilot – Focus: Type II Diabetes & CVD (~75 patients)	BHCA





- **Project Acronym: EUROGENE**
  - **Ethics**
  - **Regulations**
  - **Privacy**
  - **Validity**
  - **Code of Practice (model is HGC UK)**
  - **Utility**
    - **Common SNPs**
    - **Documented effect**
    - **Demonstrated efficacy of intervention**
    - **Gene x Environment**
- **Work with industry → best practice in genetics**



# Eurogene

1. The Eurogene Kit
2. The multilingual and highly secured portal
3. The reports to the individual

Making sense of complex data



## Issues...

- DTC vs. via Practitioner
- Lack of clear regulation throughout Europe
- Market Claims: *"We are finally able to decode our body's own manual for healthy living"*.
- Infrastructure (systems and software) to incorporate PG in healthcare



# Personal Genetics in Practice

Nutritionists, Dieticians, Medical doctors:

- Easy
- Fast
- Evidence
- Useful / effective





# Clinical decision structure

- *Research results* ... average of 17 years to be implemented in clinical practice
- *Critical component* ... centrally managed repositories of authoritative, computer-processable knowledge on how genetic and genomic interventions should be utilized clinically

## **Most significant features:**

1. Automatic ... part of clinician workflow
2. *Recommendations* not just assessments
3. Provide at the time and location of decision making

Kawamoto K, Lobach DF, Willard HF, Ginsburg GS.

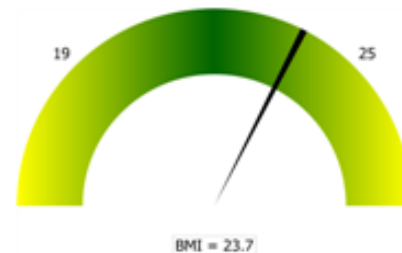
A national clinical decision support infrastructure to enable the widespread and consistent practice of genomic and personalized medicine.

*BMC Med Inform Decis Mak.* 2009 Mar 23;9:17



ID	A122334
Date of Birth	14.06.1978
Current Age	31
Gender	Male

	Normal range	Result	Status
Height		183 cm	
Weight		79.5 kg	
BMI	18.5 - 25	23.7	Normal
Waist-Circumference	Male: < 102 cm	98 cm	Normal



### Summary Profile

#### MAJOR RISK COMBINATIONS

Gene	Nutrition	Risk	Current Status	Recommendation
MTHFR CBS	Low dietary folic acid, vitamins B6 and B12	High Homocysteine levels – associated with CVD and other conditions	Homocysteine: 12 µmol/L Folate: 13 ng/ml B6: 5.2 µg/L B12: 125 µg/L	Increase the nutritional components to reach recommended goals, retest Homocysteine regularly until stable
VDR COL1A1	High caffeine Low vitamin D	Decreased bone mineral density / osteoporosis	25OH Vitamin D: 24 ng/ml Osteocalcin: 17 ng/ml	Although nutrition indicates less than adequate vitamin D intake the plasma vitamin D levels are within normal range at present. Check dietary intake and sunlight exposure and retest 25OH Vitamin D regularly to ensure stable levels

#### BIO-ANALYSIS: ANALYTE LEVELS OUTSIDE OF NORMAL RANGE

Analyte	Result	Normal Range
Total Cholesterol	356 mg/dl	< 200 mg/dl
LDL-Cholesterol	245 mg/dl	100 – 129 mg/dl
Triglycerides	180 mg/dl	< 200 mg/dl
Homocysteine	12 µmol/L	5 – 10 µmol/L

Analyse - Moz

File Edit View

Analyse

HomeAnalysis

Klienten- und P (A500140)

Persönliche Inform  
Letzte Analyse  
Alle Analysen ans

Ärztliche Anwe

Patientenliste  
Patienten anlegen  
Patienten suchen

Patient zu meiner  
Username:   
Password:

Find: aedes

Done

Deutsch

ht

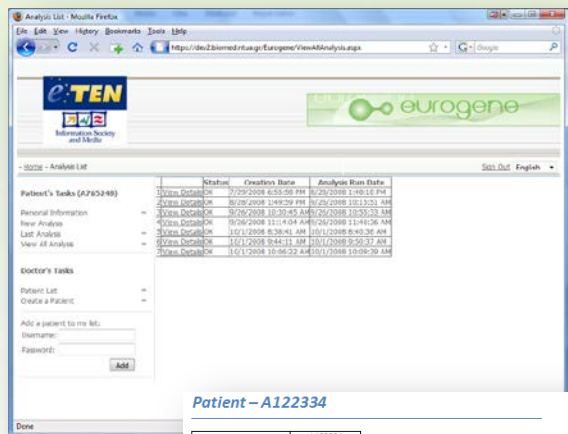
↑

↓



# Eurogene Portal

- Rules Toolset: editors, webservices, databases, secure communications etc
- Highly flexible system for creating "models" which generate the patient recommendations
- Flexible: any data (genes, diet, biomarkers, omics) in → Delivery of recommendations
- Data: secure storage, anonymous
- Continual data analysis and discovery



## Patient - A122334

ID	A122334
Date of Birth	14.06.1978
Current Age	31
Gender	Male

	Normal range	Result	Status
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VDR COL1A1	High caffeine Low vitamin D	Decreased bone mineral density / osteoporosis	25OH Vit D: 24 pp/10000000 068000460: 4.7 ng/ml	although nutrition indicates less than adequate vitamin D intake the plasma vitamin D levels are within normal range at present. Check dietary intake and sunlight exposure and retest 25OH Vit D regularly to ensure stable levels.

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# Model Editor

SafeCape Modeller v.1.0.0.0 - EuroGENE.en-US

File Quality Control Report

Model Sample Input External Input Output

EuroGENE.en-US

- Constants
- Questionnaire
- Substitution Text
- SNPs
- Risk\_Goal
- TextFormulas
- Graphs
- AddressLabel
- POR
- Biomarkers
  - Biomarker\_formulas
    - Total\_chol\_level [Too high]
    - LDL\_chol\_level [Too high]
    - HDL\_chol\_level [Normal]
    - TG\_level [Normal]
    - Waist\_goal [< 0.88]
    - SatFat\_recc [Reducing saturated fat and triglycerides in the diet]**
    - PhysAct\_recc
    - Omega3\_recc
    - OliveOil\_recc
    - LDL\_graph\_mark\_1 []
    - LDL\_graph\_mark\_2 []
    - LDL\_graph\_mark\_3 []
    - LDL\_graph\_mark\_4 []
    - LDL\_graph\_mark\_5 []
    - LDL\_graph\_mark\_6 []
    - LDL\_graph\_mark\_7 []
    - LDL\_graph\_mark\_8 []
    - LDL\_graph\_mark\_9 []
    - LDL\_graph\_mark\_10 []
  - Biomarker\_text

**General**

Description	
Handle Text	
Name	SatFat_recc
Type	Formula
Visibility	inherit (published)

**Misc**

ChkAll	True
ChkResult	
Dependencies	Model.DependencyList
ExternalKey	
FullName	EuroGENE.en-US.Biomarker_formulas.SatFat_recc
Handle	normal
InvolvedVariables	Variable[] Array

Validate  Auto

Biomarker\_text.MTHFR\_677\_CC  
Biomarker\_text.MTHFR\_677\_CT  
Biomarker\_text.MTHFR\_677\_TT  
Biomarker\_text.MTHFR\_1298\_AA  
Biomarker\_text.MTHFR\_1298\_AC  
Biomarker\_text.MTHFR\_1298\_CC  
Biomarker\_text.MS\_MTRR\_66\_AA  
Biomarker\_text.MS\_MTRR\_66\_AG  
Biomarker\_text.MS\_MTRR\_66\_GG  
Biomarker\_text.MTR\_2756\_AA  
Biomarker\_text.MTR\_2756\_AG  
Biomarker\_text.MTR\_2756\_GG  
Biomarker\_text.CBS\_699\_CC  
Biomarker\_text.CBS\_699\_CT  
Biomarker\_text.CBS\_699\_TT  
text.Sat\_Fat\_Above\_risk  
text.Sat\_Fat\_Above\_norisk  
text.Sat\_Fat\_at\_below\_risk  
text.Sat\_Fat\_at\_below\_norisk  
text.OliveOil\_above\_risk  
text.Omega3\_above\_risk  
text.Omega3\_above\_male\_norisk  
text.Omega3\_above\_female\_norisk  
text.Omega3\_at\_risk  
text.Omega3\_at\_male\_norisk  
text.Omega3\_at\_female\_norisk  
text.Omega3\_below\_risk  
text.Omega3\_below\_male\_norisk  
text.Omega3\_below\_female\_norisk  
text.exercise\_above\_risk  
text.exercise\_above\_norisk  
text.exercise\_at\_risk  
text.exercise\_at\_norisk  
text.exercise\_below\_risk  
text.exercise\_below\_norisk  
Biomarker\_text.smoking\_above  
Biomarker\_text.smoking\_below  
Illegal name characters

Formula Editor - Biomarker\_formulas.SatFat\_recc

```

1 if ( ( Risk_Goal.Sat_Fat.Risk = "True" ) and ( Risk_Goal.Sat_F
2 Biomarker_text.SatFat_reduce
3 } else {
4   ""
5 }

```

Result: Reducing saturated fat and triglycerides in the diet

```

if ( ( Risk_Goal.Sat_Fat.Risk = "True" ) and ( Risk_Goal.Sat_Fat.Th
Biomarker_Text.SatFat_reduce
} else {
""
}

```

Name


Ready

# Report template editor

Eurogene\_Biomarker\_V2sample.docx - Microsoft Word

Table Tools

Home Insert Page Layout References Mailings Review View Acrobat Report Editor Design Layout



BMI = 23.7

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VDR COL1A1	High caffeine Low vitamin D	Decreased bone mineral density / osteoporosis	25OH Vit D: 24 ng/ml Osteocalcin: 17 ng/ml	VitaminD_5

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Triglycerides	180 mg/dl	< 200 mg/dl
Homocysteine	12 µmol/L	5 – 10 µmol/L
Vitamin B12	125 µg/L	200 – 1000 µg/L

Report Editor

Load Model

Add Version Tag Combined Version Tag

Bindings Model Formulas

Bind Add Graph Bind All

- PON1\_Leu55Met [Yes]
- PPARg [Yes]
- MnSOD\_C\_28T [Yes]
- SOD3\_C760G [No]
- TNFA\_G\_308A [No]
- VDR\_Bsm [Yes]
- VDR\_Taq [Yes]
- VDR\_FokI [No]
- Antioxidants
- VitD\_Calcium
  - VitaminD\_1
  - VitaminD\_2
  - VitaminD\_3
  - VitaminD\_4
  - VitaminD\_5 [Based on the [VDR] variations in your genes]
  - VitaminD\_6
  - VitaminD\_7
  - VitaminD\_8
  - Calcium\_1
  - Calcium\_2
  - Calcium\_3
  - Calcium\_4
  - Calcium\_5 [Based on your genetic variations in the [VDR]]
  - Calcium\_6
  - Calcium\_7
  - Calcium\_8
  - Vit\_D\_toxic [Important Note: Based on your questionnaire]
- Chrom\_Salt
- Macronutrients
- Lifestyle
- Graphs
- AddressLabel
- POR
- Biomarker formulas

Page: 1 of 5 Words: 690 English (United Kingdom) 100%

# Regulations



European Journal of Human Genetics (2011) 19, 382–388  
© 2011 Macmillan Publishers Limited All rights reserved 1018-4813/11  
[www.nature.com/ejhg](http://www.nature.com/ejhg)

## ARTICLE

# Personal genetics: regulatory framework in Europe from a service provider's perspective

Keith A Grimaldi<sup>\*1</sup>, Markus P Look<sup>2</sup>, G Antonio Scioli<sup>3</sup>, Juan Coll Clavero<sup>4</sup>, Stathis Marinos<sup>5</sup> and Tassos Tagaris<sup>1</sup>



# Marketing

"...use *Genetics* as the credibility tool ... to lose weight. Focus on the initial naivety of the individual in believing in the way things work. Similar to most gimmicky weight loss marketing programs."

Email from XXXX to me



# Weight loss?

Clinically shown to improve weight loss by over 2.5

to help you lose properly matching genotypes. No g

Now you can o  
supplem



Peso forma  
per sempre

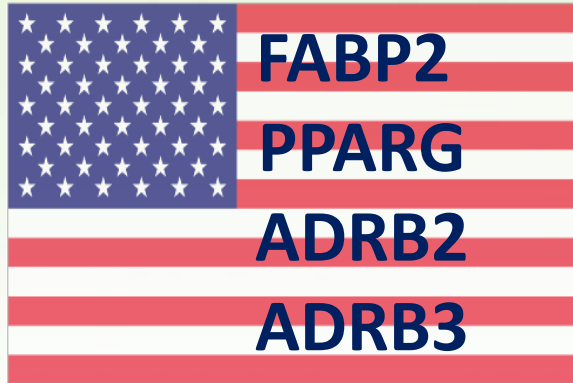
Personalizza  
la tua alimentazione,  
raggiungi il peso forma  
e mantienilo per sempre.

for your genes.  
DNA will be thrilled!



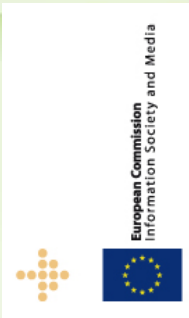


# Nutrigenetics & weight loss?



Low calorie, low carb, high carb, low fat, etc... according to your genes???





## Code of Practice

- Claims must be accurate
- Evidence & methods transparent
- Scientific validation
- Privacy and use of customer's DNA
- Full and clear information for the customer to understand the test including accuracy and limitations
- Recommendations to purchase follow-on products/services (e.g. diets, supplements) must be fully supported by scientific evidence



# Collaborations



Spin off  
[www.safecape.gr](http://www.safecape.gr)



Neonatal – CD and LCT



NTUA  
Euthenics – Greek sponsored NewCo



Nutrigenomics in South Africa  
<http://www.dnadiet.co.za/>



# Collaborations



[www.iabc.ch](http://www.iabc.ch)

Biomarkers & genetics



NewCo - Canada



Personalised genetics – DTC, Italy

**66° CONGRESSO FIMMG-METIS**



Nutrigenomics , Italy

“La medicina generale nel terzo millennio”

dal 3 all'8 ottobre 2011

Villasimius (CA) - Tanka Village





UCD Institute of Food & Health  
[www.ucd.ie/foodandhealth](http://www.ucd.ie/foodandhealth)

# Food4me.org



**Food4me.org** is an international consortium of EU and US researchers with an interest in the subject of personalised nutrition. This consortium is actively seeking research funds for a specific set of research questions in the following areas:

- Consumer understanding of and attitudes to personalised nutrition
- Business models for the delivery of personalised nutrition
- The retail and distribution sectors
- IT solutions for self assessment of dietary habits and food choice
- Nutritional phenotyping
- Communication of personalised nutrition
- Gene-nutrient interactions

The consortium presently involves academic scientists from Dublin, Ulster, Munich, Maastricht, Reading, Lund, Zeist, Oslo Wageningen, Newcastle and Reading in the EU and from Boston and Davis in the US.

It also involves the European Union Food Information Council and two commercial organisations Nestlé & DSM.

Professor Mike Gibney from University College Dublin's Institute of Food & Health is the consortium convenor and can be contacted at

**[mike.gibney@ucd.ie](mailto:mike.gibney@ucd.ie)**





**Proposal full title: European Research on DevelopmentAL, BirTH and Genetic Determinants of Ageing**

**Proposal acronym: EurHEALTHAgeing**

**Proposal duration: 36 months**

**Type of funding scheme: Collaborative Project (Small or medium-scale focused research project)**

**Work programme topics addressed: HEALTH.2011.2.2.2-2: Linking human development and ageing.**

**Name of the coordinating person: Prof Tim Spector**

