



FishMed-PhD Teaching week

February 27th, 2026



# ***Human population dynamics in traditional fishing communities: genetics, nutrition, health***



Photo by Andrea De Giovanni

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# Summary



- Traditional fishing communities as evolutionary “natural laboratories”
- Marine resources from past to present
- Fishing: health benefits and contaminants
- Genetics and epigenetics of fishing communities

# Traditional fishing communities

## Who are they?

Traditional fishing communities are **groups with deep-rooted cultural, economic, and social ties to the sea**, relying on intergenerational knowledge for sustainable, small-scale fishing.

They serve as stewards of marine ecosystems, often employing eco-friendly techniques, but face critical threats from industrial overfishing, environmental degradation, and climate change.



Coastal fishing communities are unified through fishing.

Source: ([Vietnamese Private Tours on Pexels, 2018](#))

## Key aspects of traditional fishing communities include:

***Cultural Identity and Heritage:*** Fishing is not just an occupation, but a lifestyle, often accompanied by rituals and deep spiritual connections to the sea.

***Sustainability and Knowledge:*** These communities often use selective, traditional, or biodegradable gear and possess rich, experiential knowledge of local marine ecosystems.

***Social Structure:*** They are often organized around cooperative, familial, and community-based structures, with experienced fishers or elders often guiding community decisions.

***Challenges:*** They face significant pressure from large-scale commercial fishing, coastal development, pollution, and climate change, which disrupts fish migration and reduces stock.

***Role in Conservation:*** Despite challenges, they play a crucial role in maintaining biodiversity and regulating local fishing grounds through traditional management practices.

These communities are often at the "crossroads" of modernization, fighting to maintain their traditions while battling economic marginalization and environmental changes

# Traditional Fishing Community

FS is a defined group of people **who share identity and** interact each other to perform activities along the **fisheries value chain based on experiential knowledge accumulated over time and passed along generations.**



Traditions represent a set of practices developed by repetition over time and regulated by shared values, beliefs, accepted rules, and negotiated across different levels and consist of the cultural background and identity of specific groups of people who exercise them. **Traditional usually refers to cultural continuity transmitted in the form of social attitudes, beliefs, principles, and conventions of behavior and practice derived from historical experience.**

Traditional fishing communities—including **Arctic Inuit, Mediterranean coastal populations, Pacific Islanders, and Southeast Asian littoral societies**—occupy ecological niches characterized by high reliance on marine resources.

These populations frequently demonstrate distinctive **demographic structures, dietary compositions, and health profiles shaped by long-term environmental exposure.**

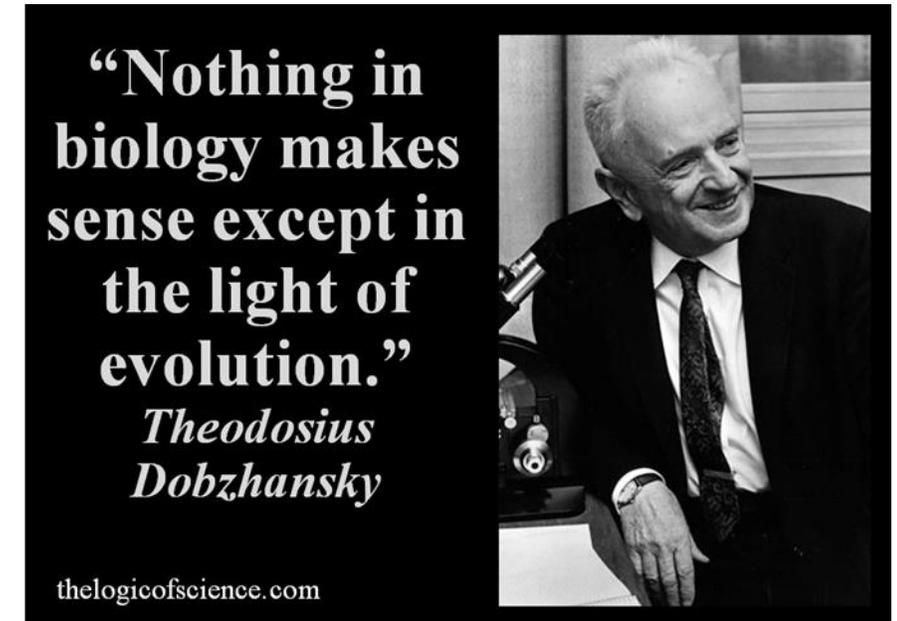
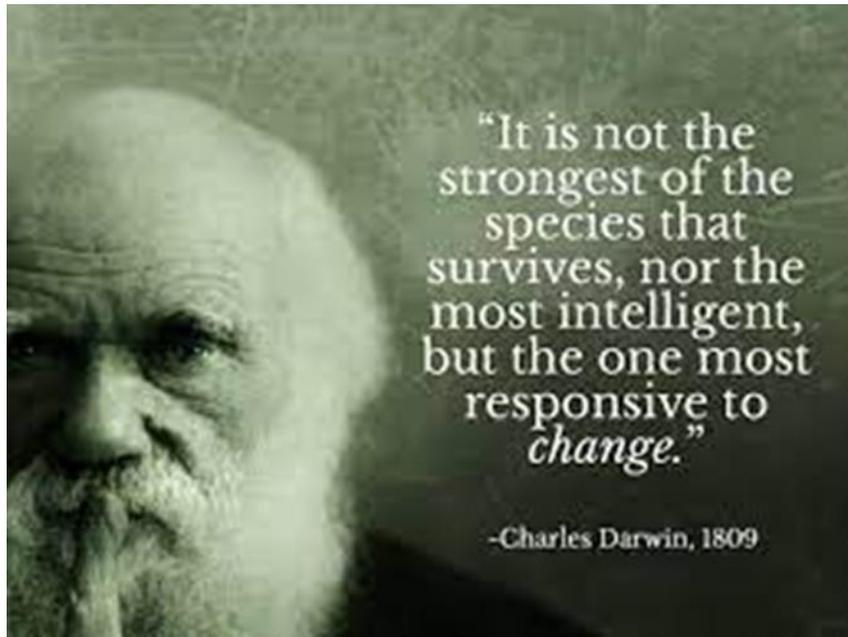


Traditional fishing communities represent **unique biocultural systems** in which ecological constraints, marine-based subsistence, demographic structure, and genetic processes intersect. These populations offer valuable insight into **microevolutionary dynamics, dietary adaptation, and the health consequences of rapid modernization.**



From **an evolutionary perspective**, fishing communities provide valuable case studies for examining gene–environment interaction, microevolution in small effective populations, and nutritional adaptation under ecological constraints.

The increasing pace of modernization and climate change now challenges these historically stable adaptive systems.



These populations frequently demonstrate **distinctive demographic structures** (small effective population sizes, historical isolation, maritime trade networks), dietary compositions (high protein and omega-3 fatty acid intake), and health profiles.

From an evolutionary perspective, such communities serve as natural laboratories for examining:

- Microevolutionary processes in small populations
- Gene–environment interaction
- Nutritional adaptation and mismatch
- The interaction between ecological stability and epidemiological change

Understanding these systems is increasingly urgent as globalization, climate change, and dietary transition alter the ecological contexts that shaped human adaptation over millennia.

# HOW TO DEFINE A HUMAN POPULATION:

**Demography:** Focuses on the number, structure, and distribution of individuals.

**Biology:** Defines populations from a genetic perspective.

A population is the basic unit of biological evolution. A "Mendelian population" (deme) is:

1. A reproductive group of interbreeding individuals living together, and
2. Sharing a common gene pool (the set of genes carried by individuals in the population).

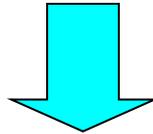
## **Population as a Spatio-Temporal Group:**

Individuals from different age groups do not belong to the same reproductive group, even if they live in the same area and share a genetic pool.

# What is a population?

Before defining an allele's freq. within a population,  
a population concept must be defined

## Breeding Population



**A group of individuals within a species  
who are reproductively isolated from other groups...**

they restrict mate selection to members of their own group  
usually due to geographical and/or social barriers that prevent  
mating with outsiders

# Evolutionary & Genetic Foundations of Marine-Dependent Populations

Why fishing communities are evolutionary “**natural laboratories**”?

- Small  $N_e$ , ecological constraints, gene–culture coevolution
- Isolation and gene flow
- Founder effect and genetic drift
- Impact of trade and mobility
- A biocultural framework

(a holistic approach that treats biological ecosystems and human cultural systems as inextricably linked and co-evolved. It moves beyond traditional "bio-centric" management by recognizing that protecting fish stocks also requires protecting the traditional knowledge, rights, and identities of the people who harvest them!)

# HARDY-WEINBERG EQUILIBRIUM

## CONDITIONS:

- Diploid organism, sexual reproduction
- Non-overlapping generations
- Random mating
- Large population size
- Negligible mutation rate
- Negligible migration rate
- Mortality independent of genotype
- Fertility independent of genotype

## IF THESE CONDITIONS AREN'T

### MET:

- Random mating
- Large population size
- Negligible mutation rate
- Negligible migration rate
- Mortality independent of genotype
- Fertility independent of genotype

**INBREEDING**

**GENETIC DRIFT**

**MUTATION**

**MIGRATION**

**SELECTION**

**SELECTION**



# Population Structure and Microevolutionary Dynamics

## Effective Population Size and Genetic Drift

Effective population size ( $N_e$ ) is often substantially smaller than census size ( $N_c$ ) in small-scale fishing communities due to:

- Endogamy
- Unequal reproductive success
- Seasonal male mortality (historically)
- Migration bottlenecks

A migration bottleneck refers to a, usually, ***geographic or environmental constraint*** that forces migrating populations into a narrow, concentrated area, drastically limiting their movement and increasing vulnerability.

## •Inbreeding

- Inbreeding**: is the mating of relatives - a form of non-random mating.

- Inbreeding**: alone does not change allele frequencies, but inbreeding *does* change genotype frequencies.

- The main effect of inbreeding** is to increase the frequency of homozygous genotypes in a population (main result: Excess Homozygotes), relative to the frequency that would be expected with random mating.

- ★ **Unlike random mating**, which may affect some genes but not others, inbreeding affects all genes in the genome.

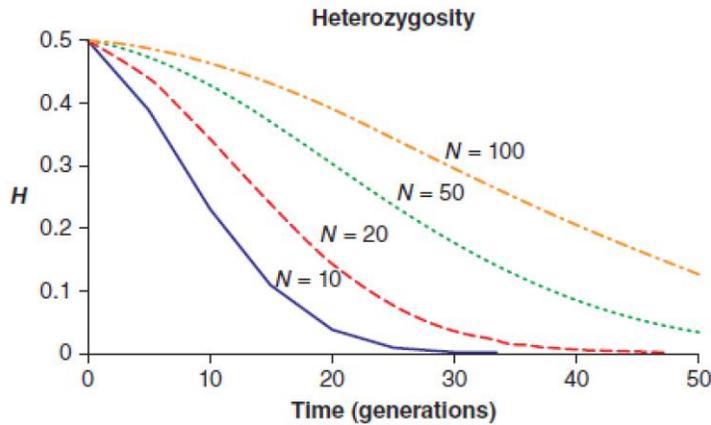
# Ne=effective population size

The size of the breeding population (**the effective population size**) governs the strength of the drift effect.

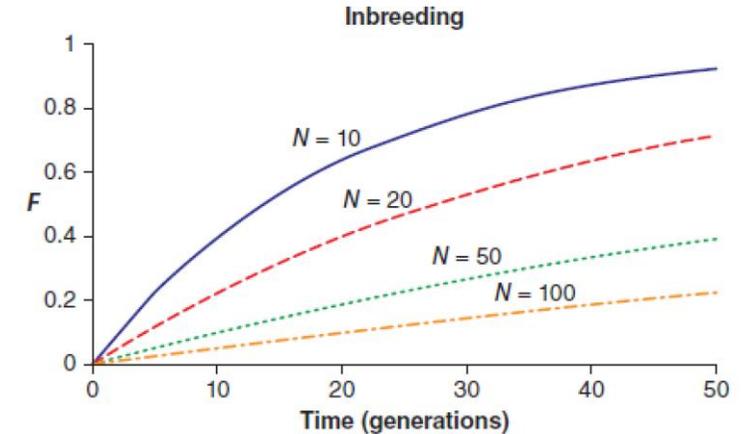
When the effective population size is small, genetic drift will be stronger.

## Drifting alleles usually have a finite lifetime.

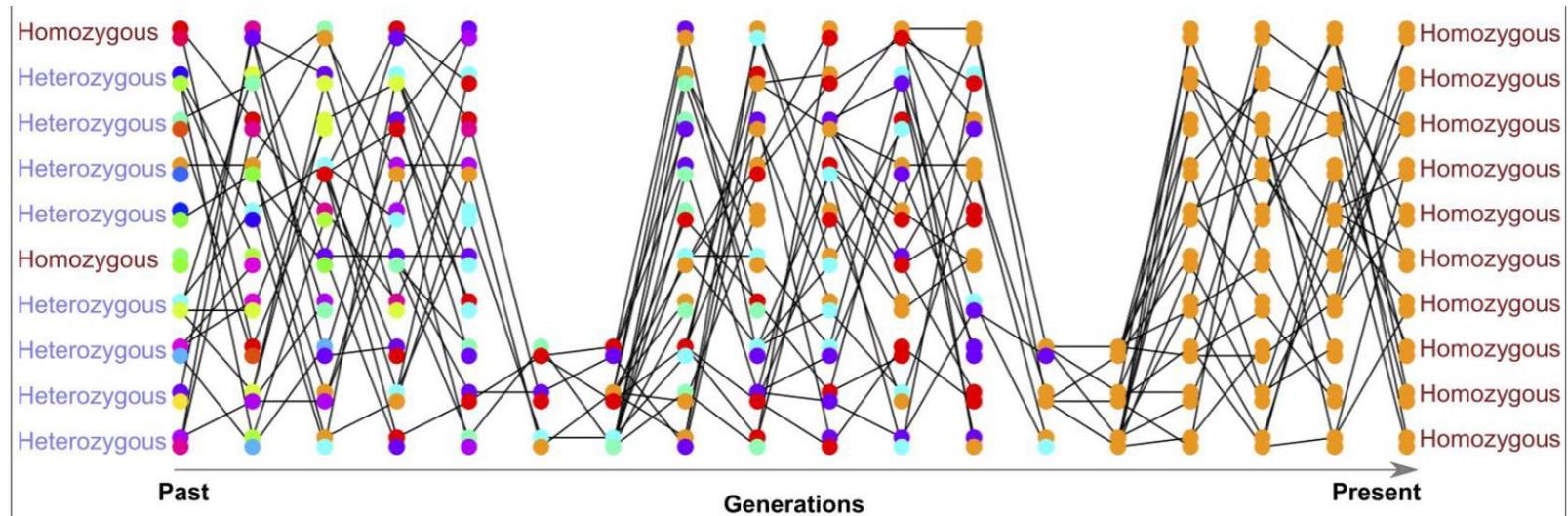
As the frequency of an allele drifts **up and down over successive generations**, eventually it drifts until fixation - that is, it either reaches a frequency of zero, and disappears from the population, or it reaches a frequency of 100% and becomes the only allele in the population.



**FIGURE 5.3**  
Loss of heterozygosity ( $H$ ) over time as a function of population size ( $N$ ).

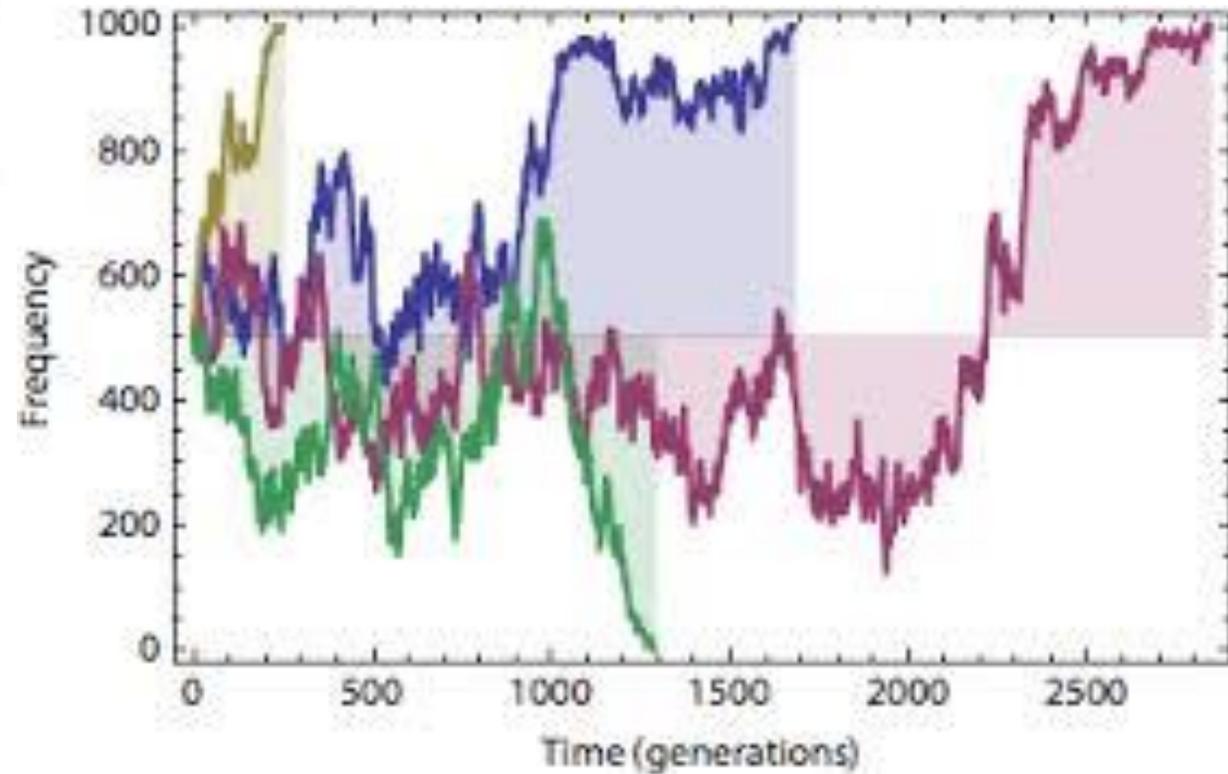
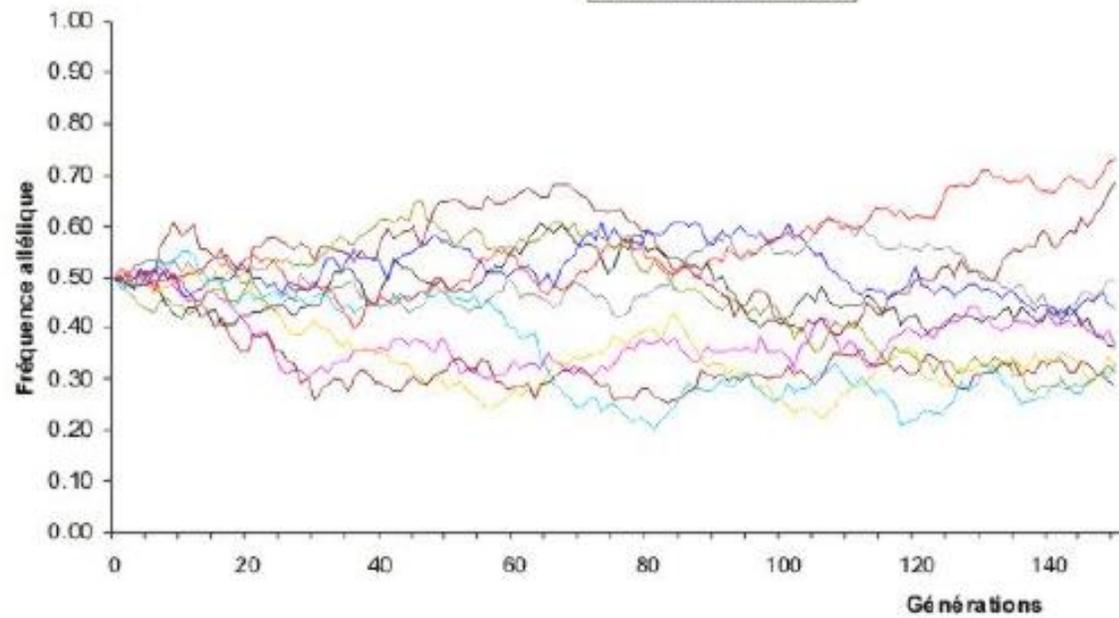


**FIGURE 5.4**  
Increase in inbreeding ( $F$ ) over time as a function of population size ( $N$ ).

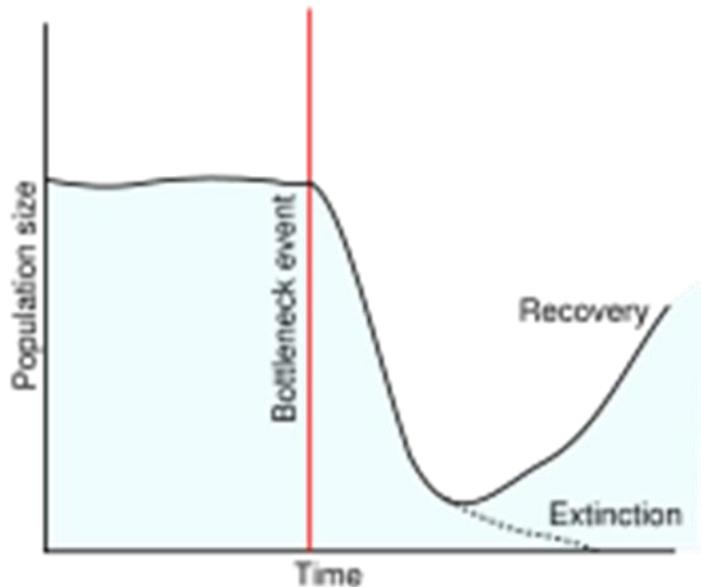


## Rate of drift is stronger in small populations

10 replicates  $2N=1000$

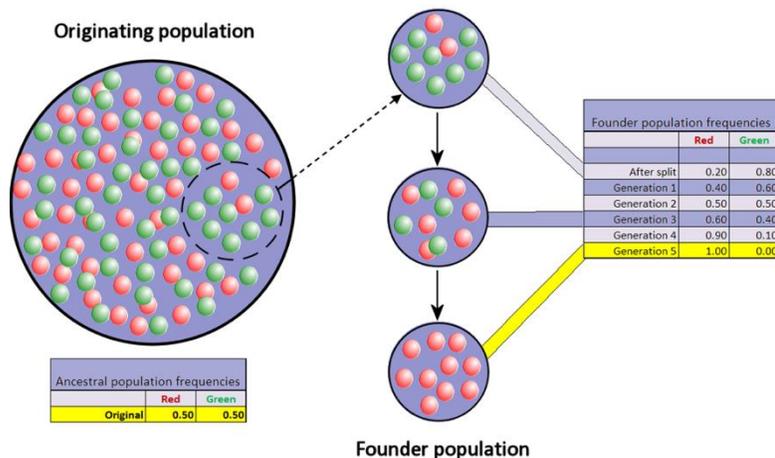


In populations where  $N_e < 500$ , drift can substantially alter allele frequencies within relatively few generations. This has implications for interpreting putative adaptive signals: allele frequency shifts may reflect stochastic processes rather than directional selection.



## Founder Effects and Maritime Colonization

Island colonization and Arctic settlement frequently involved **serial founder events**. Reduced genetic diversity and elevated linkage disequilibrium characterize such populations. This complicates selection detection methods, which must disentangle demographic history from adaptive processes.



Comparatively:

- Arctic Inuit populations demonstrate strong bottleneck signatures.
- Mediterranean fishing communities often show greater gene flow via trade routes.

## **Founder Effects and Maritime Colonization**

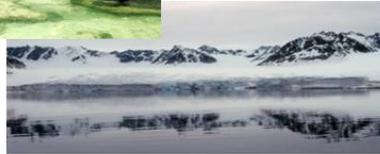
Serial founder events during Arctic settlement and island colonization reduced genetic diversity and increased linkage disequilibrium. These demographic processes complicate detection of positive selection because demographic bottlenecks can mimic selective sweeps (Nielsen et al., 2017).

## **Gene Flow in Coastal Networks**

Mediterranean fishing communities historically maintained maritime trade routes, facilitating gene flow and reducing differentiation compared to more isolated Arctic groups. Thus, demographic history must be integrated into interpretations of adaptation.

## Gene Flow Through Trade Networks

Coastal populations are paradoxically both isolated and connected. While ecological niches may limit inland admixture, maritime trade can facilitate long-distance genetic exchange. Mediterranean communities historically exhibited greater connectivity compared to Arctic groups, producing different patterns of population structure (e.g., lower FST among Mediterranean subpopulations).



**Colonization** = movement into previously unoccupied land



***Founder effect***

**Migration** = movement into an occupied land



***Gene flow***

(migrants contribute to the next generation in their new location)



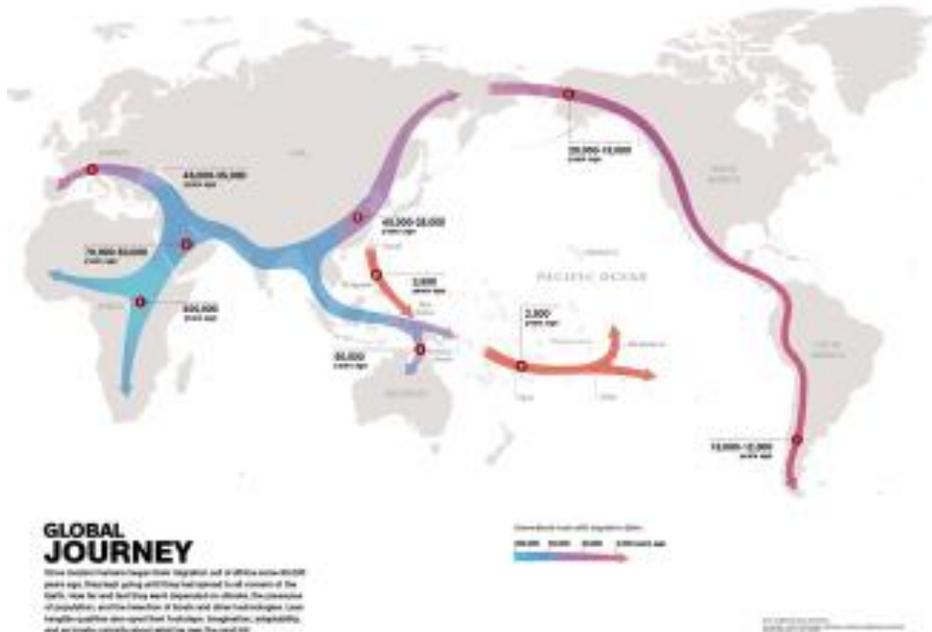
# Summary



- Traditional fishing communities as evolutionary “natural laboratories”
- **Marine resources from past to present**
- Fishing: health benefits and contaminants
- Genetics and epigenetics of fishing communities

# Marine resources from past to present

Why does the fish story from our evolutionary past matter today?

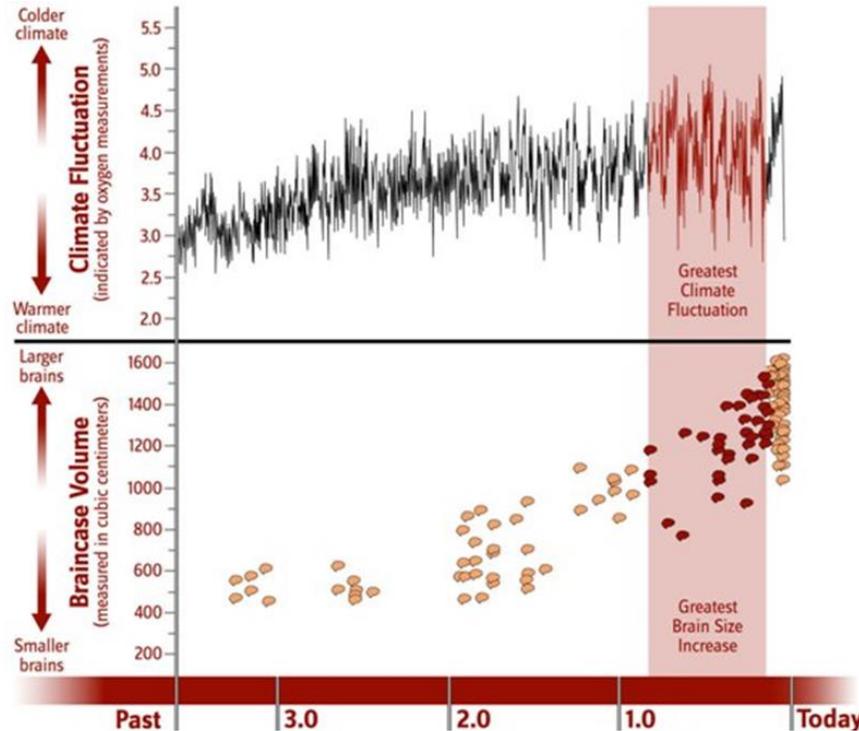


**Coastal adaptations** have become an important topic in discussions about the evolution and dispersal of *Homo sapiens*.

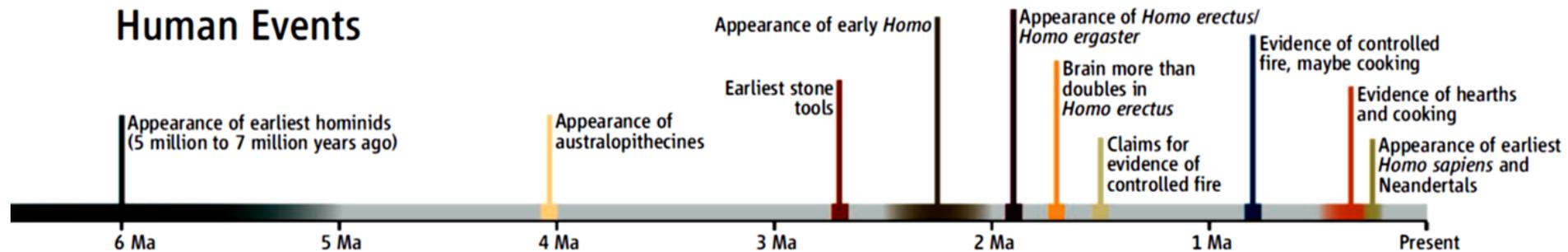
# The 'brain hypothesis' for the causal impact of coastal adaptations for human evolution



Over the course of human evolution, brain size tripled



The human brain is nearly 60% fat by total weight, and that big, **powerful brain needs to be provided with certain types of fats (both saturated and unsaturated)** throughout life to provide a balance of structural integrity and fluidity to its cells.

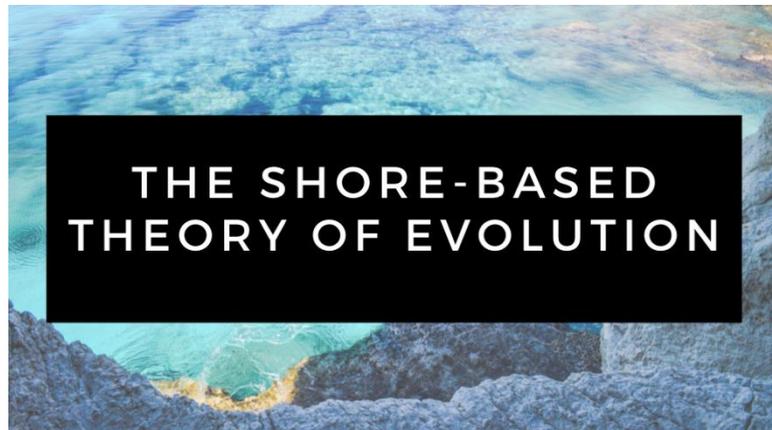


# What are the selective advantages conferred by adopting coastal adaptations?

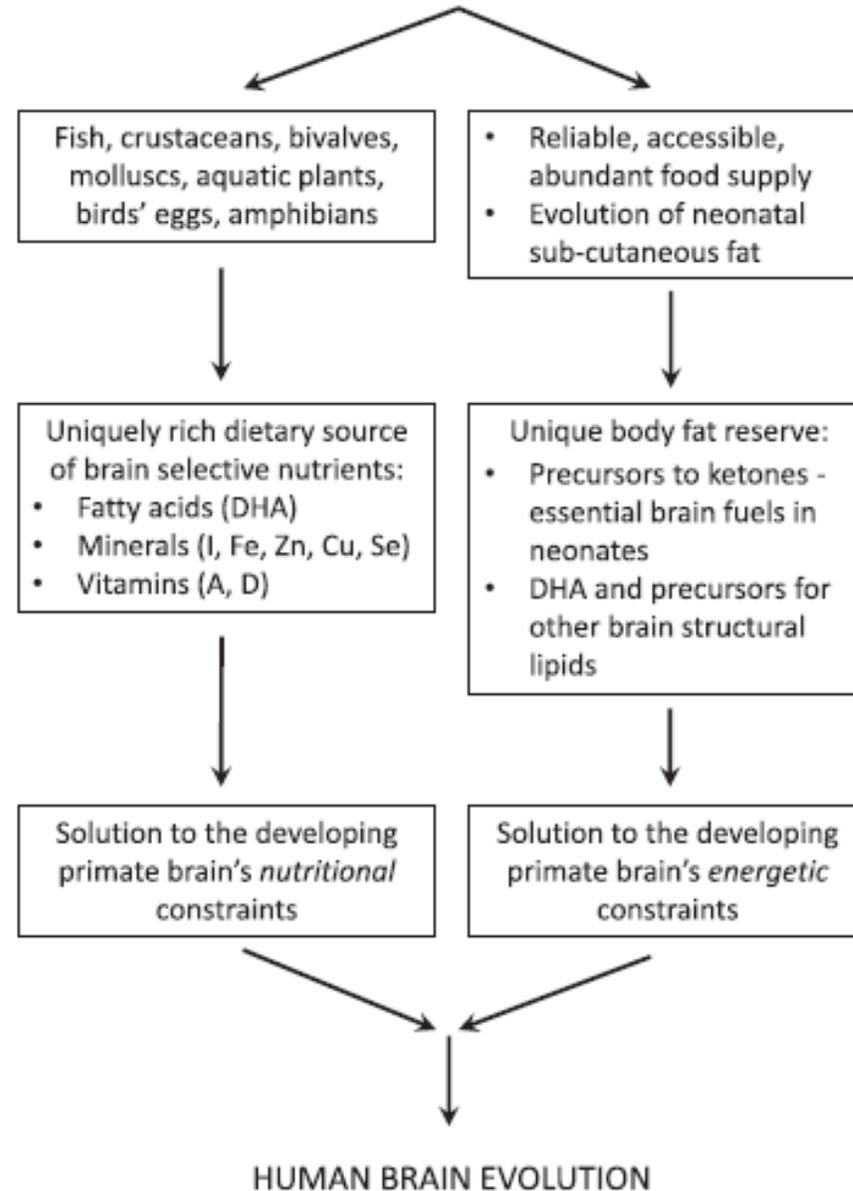
**Shore-based habitats** provided abundant and sustained access to a wide selection of foods rich in brain selective nutrients



associated to the evolution of neonatal body fat reserves, which were just as important for optimal human brain development.



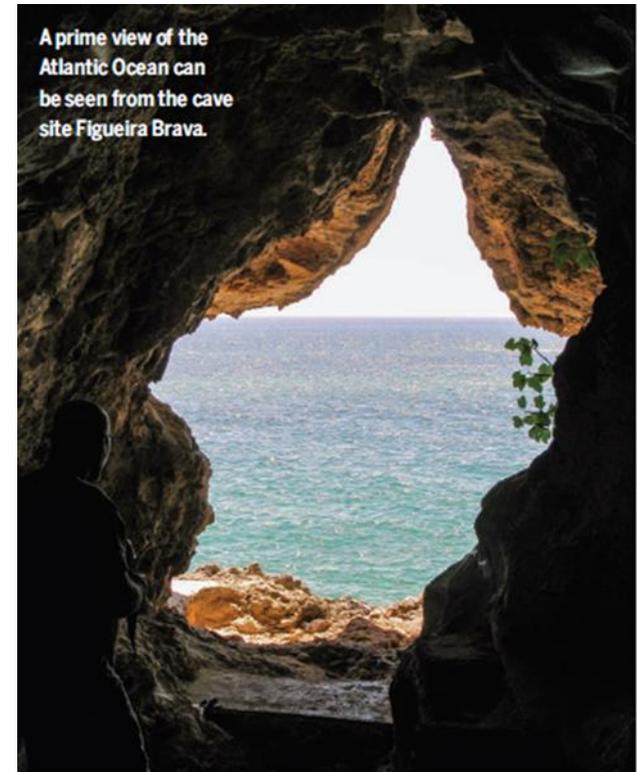
## THE SHORE-BASED PARADIGM: Solution to two major constraints on brain evolution



# Did Neanderthal adapt to the sea in the same way as early *Homo sapiens*?



Archaeological research in southern Africa revealed early human coastal adaptations that occurred at least as far back as **~160,000 years** ago in the Middle Stone Age (MSA)—the cultural period of the earliest *Homo sapiens*. **Paleolithic sites across Africa and elsewhere support the hypothesis that coastal adaptations have a long and lasting history.**



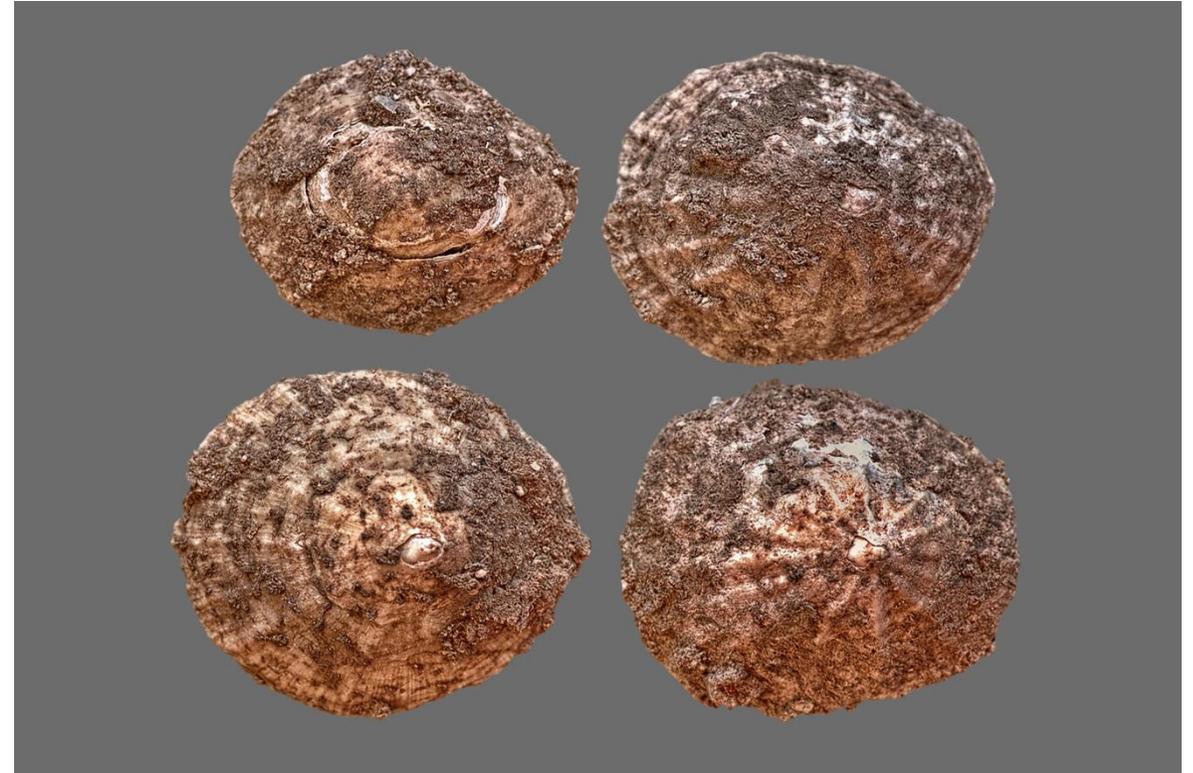
# INSIGHTS

PERSPECTIVES

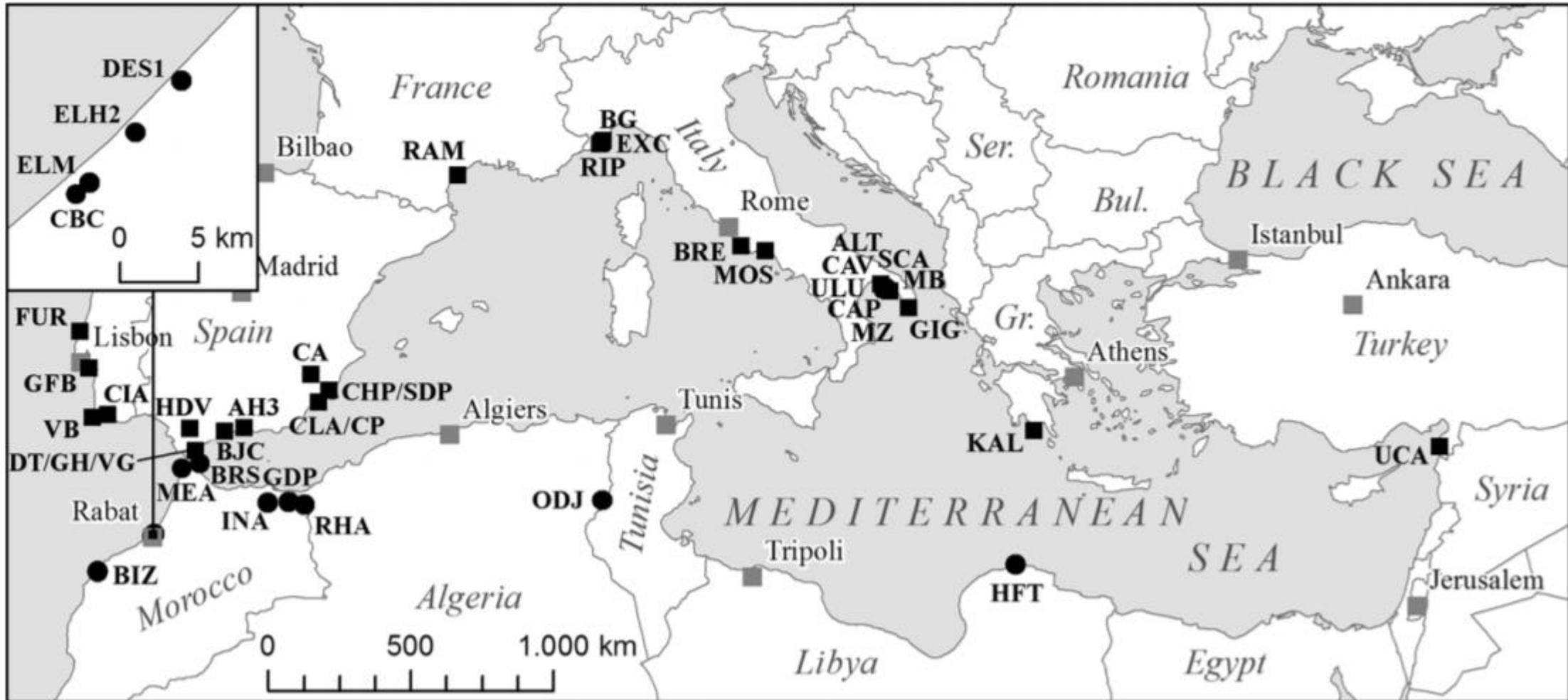
ANTHROPOLOGY

## Neanderthal surf and turf

Did our closest ancestors adapt to the sea in the same way as early *Homo sapiens*?



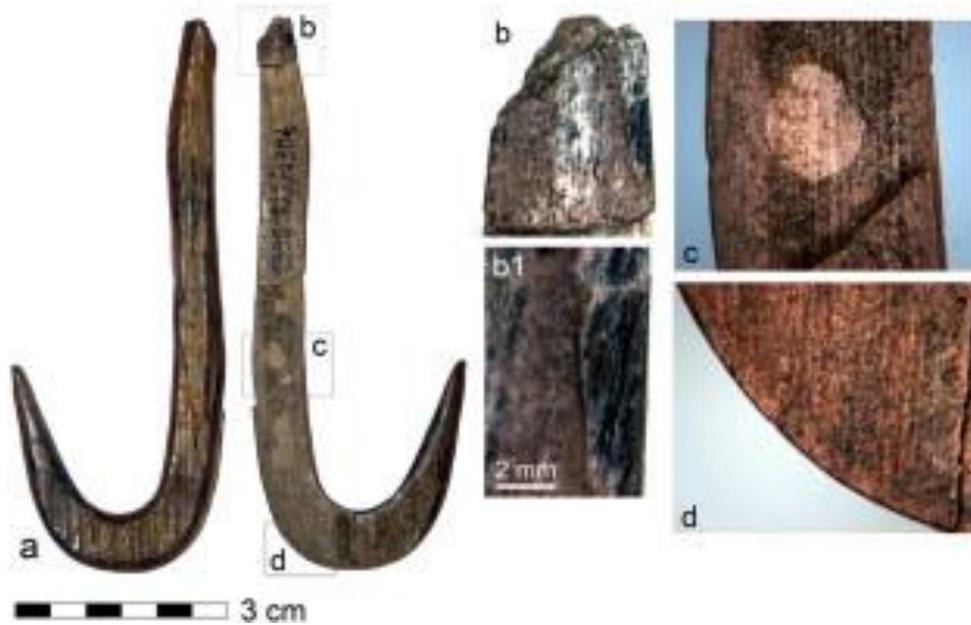
Shells of *Patella vulgata*, or common limpet, a type of edible sea snail, recovered from a seaside cave in Portugal that was once inhabited by Neanderthals. Zilhao et al. *Science*, 2020



Map depicting the main sites with evidence of coastal adaptations by modern humans in north Africa (circles) and Neanderthals in Europe (rectangles).

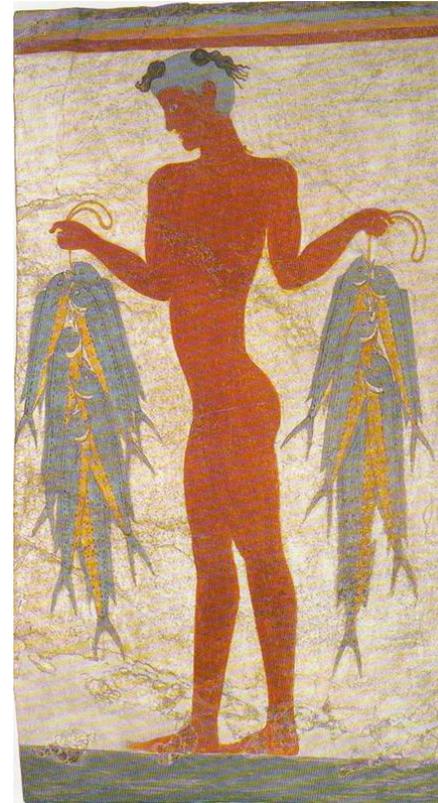
There is good evidence for Neanderthal use of marine resources and coastal landscapes from 30 cave, rockshelter and open-air sites associated with MP archaeology.

# Fishing is a prehistoric practice dating back at least 40,000 years.



An ivory fishhook with a raw material age of about 19,000 years Final Palaeolithic, site Wustermark 22 (northeastern Germany)

► Mesolithic fishhook tradition has its roots in the Final Palaeolithic.



Little Fisherman. Wall painting, Akrotiri, Thera, Greece



Aegean Talismanic seal with crab, ca. 1450 BC

# Neanderthals Consumed Crabs, Other Shellfish Over 90,000 Years Ago as Evident on Cave Remains Found in Portugal

Feb 08, 2023

Many of the shells and bones were marked with black burns, which showed they were heated to over 300°C (572°F), indicating they had been roasted for the purpose of eating.



# Archaeology Meets Marine Ecology: The Antiquity of Maritime Cultures and Human Impacts on Marine Fisheries and Ecosystems

Jon M. Erlandson<sup>1</sup> and Torben C. Rick<sup>2</sup>

Annu. Rev. Mar. Sci. 2010. 2:231–51

Understanding the antiquity of coastal settlement, marine fishing, and maritime migrations is difficult because global sea levels have risen ~125 m in the past 20,000 years, drowning ancient shorelines and vast coastal landscapes. Recent efforts to better understand the importance of marine and freshwater ecosystems in human evolution have focused on different approaches and perspectives on the **antiquity of fishing societies, seafaring, and human impacts on marine ecosystems.**

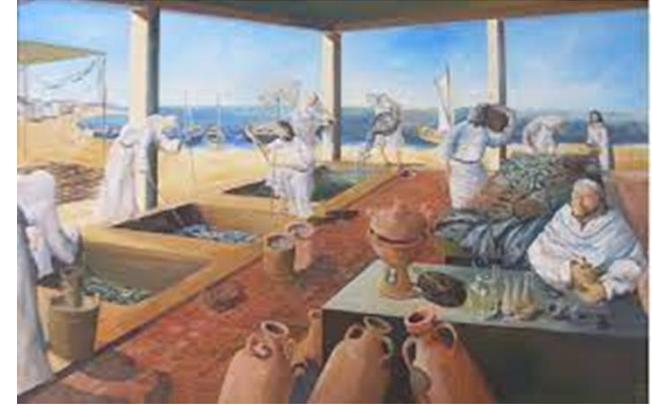
## REVIEW

# Ancient DNA Research in Maritime and Underwater Archaeology: Pitfalls, Promise, and Future Directions

Lisa Briggs



Underwater archaeologist Lisa Briggs recovering artefacts from the Queen Anne's Revenge shipwreck (Photo credit: W. Welsh)



Cytosine deamination involves the addition of an oxygen molecule and bacterial diagenesis will be significantly slowed in an anoxic environment. Moreover the 'salt effect' significantly slowed the process of depurination in high ionic environments



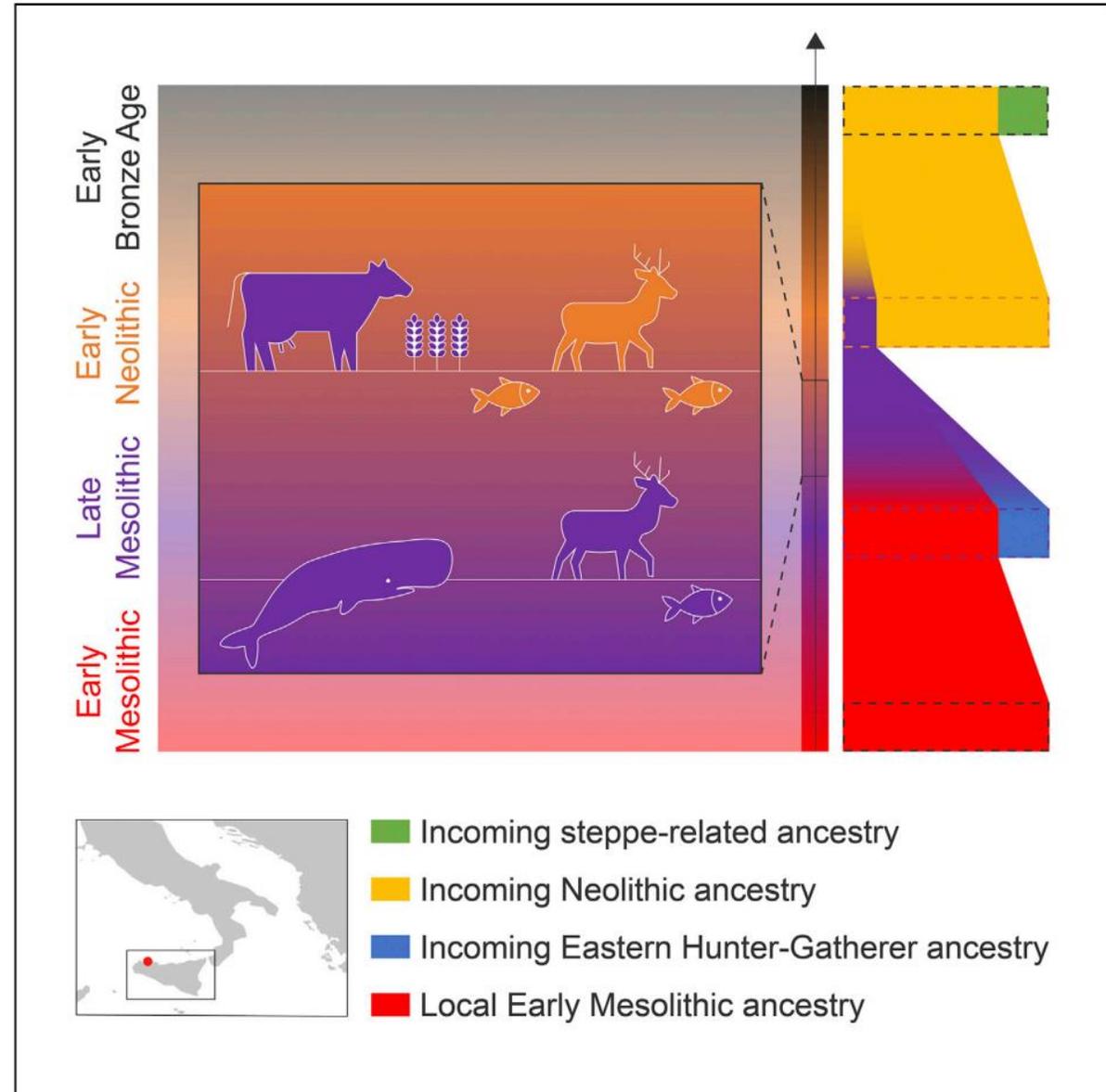
## Article

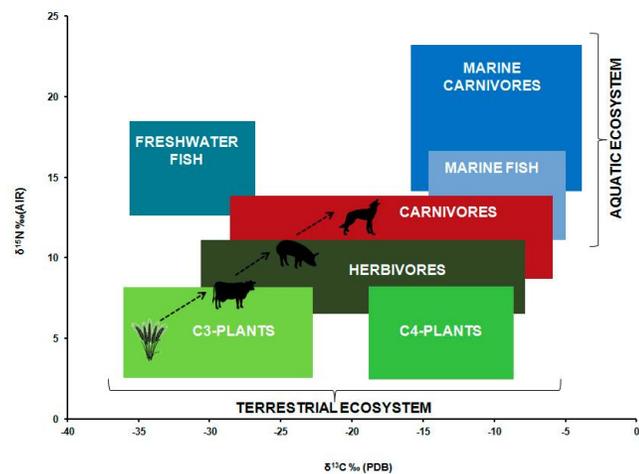
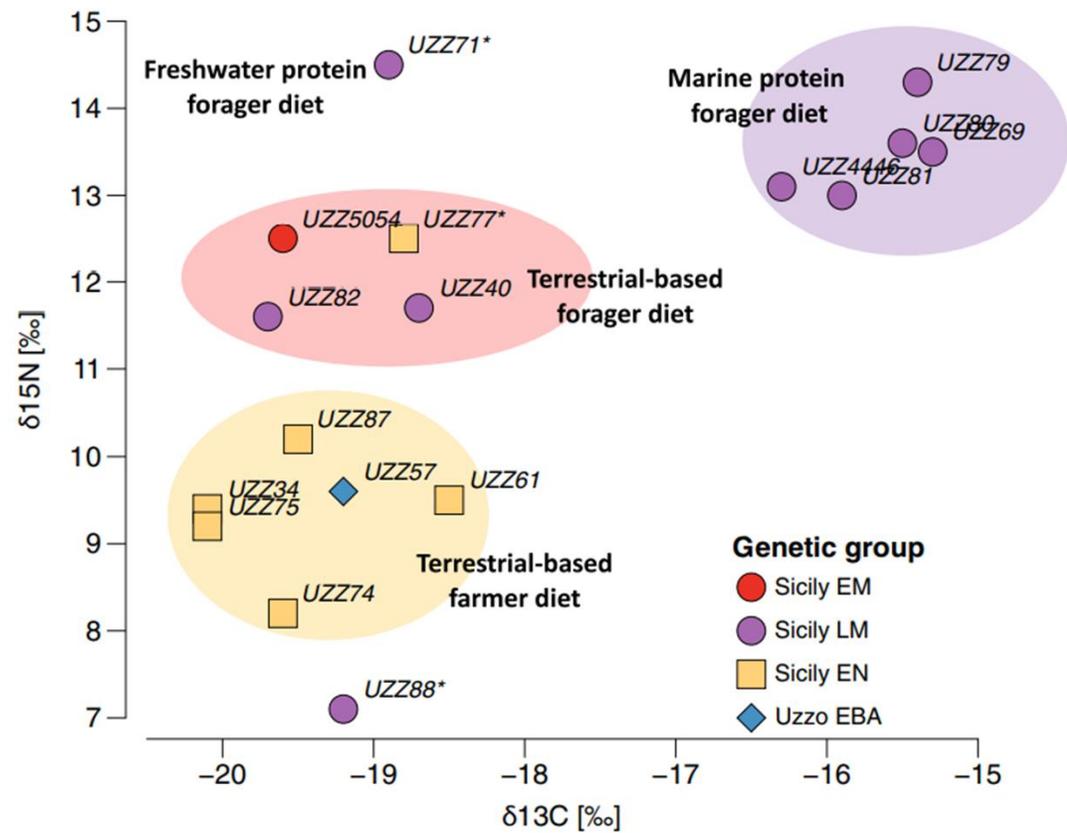
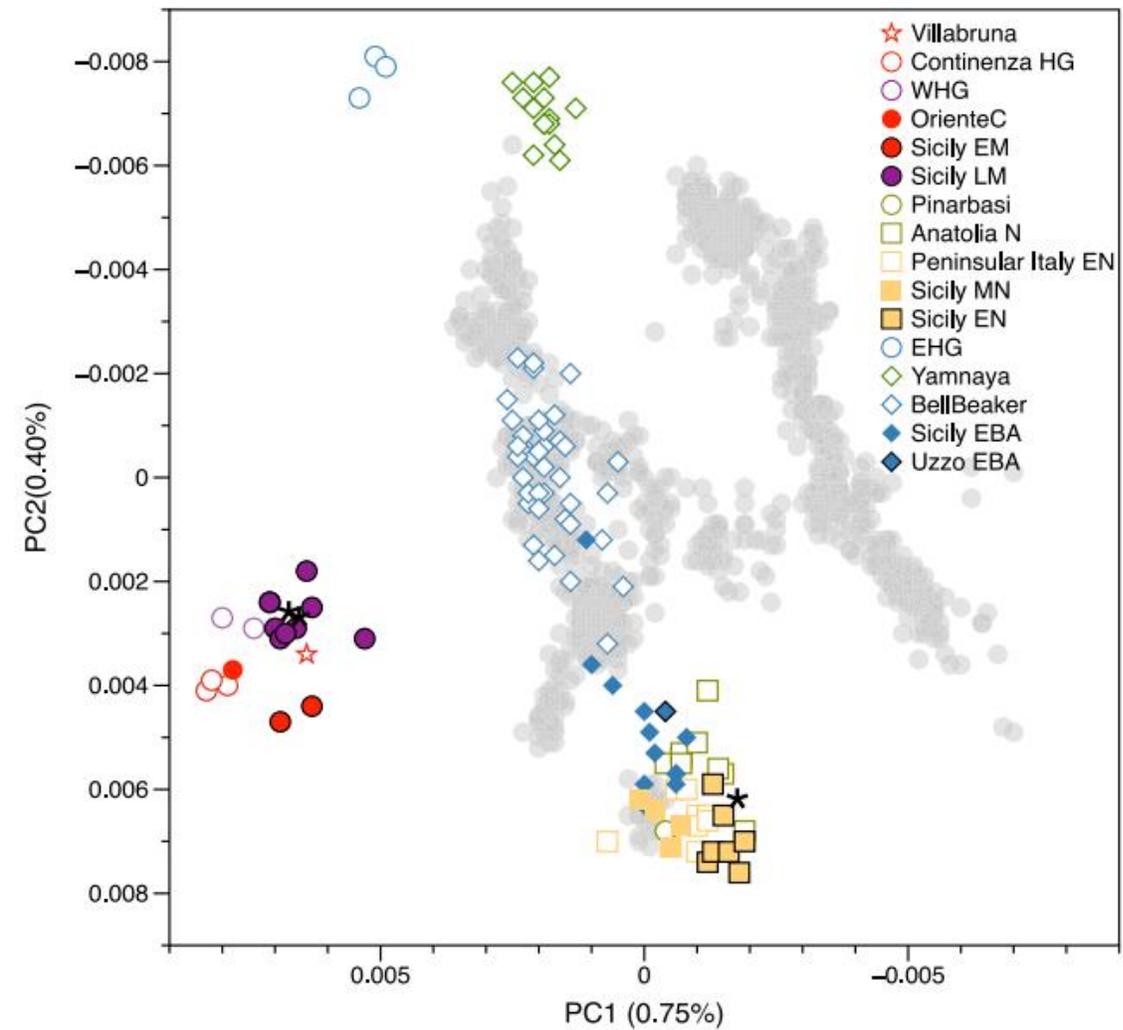
## Genomic and dietary discontinuities during the Mesolithic and Neolithic in Sicily

He Yu,<sup>1,19,\*</sup> Marieke S. van de Loosdrecht,<sup>1,19</sup> Marcello A. Mannino,<sup>2,3,19,\*</sup> Sahra Talamo,<sup>3,4</sup> Adam B. Rohrlach,<sup>1,5</sup> Ainash Childebayeva,<sup>1</sup> Vanessa Villalba-Mouco,<sup>1,6</sup> Franziska Aron,<sup>1</sup> Guido Brandt,<sup>1</sup> Marta Burri,<sup>1</sup> Cäcilia Freund,<sup>1</sup> Rita Radzeviciute,<sup>1</sup> Raphaela Stahl,<sup>1</sup> Antje Wissgott,<sup>1</sup> Helen Fewlass,<sup>3</sup> Antonio Tagliacozzo,<sup>7</sup> Marcello Piperno,<sup>8</sup> Sebastiano Tusa,<sup>9</sup> Carmine Collina,<sup>10</sup> Vittoria Schimmenti,<sup>11</sup> Rosaria Di Salvo,<sup>12</sup> Kay Prüfer,<sup>1,13</sup> Cosimo Posth,<sup>1,14,15</sup> Jean-Jacques Hublin,<sup>3,12</sup> Detlef Gronenborn,<sup>16</sup> Didier Binder,<sup>17</sup> Choongwon Jeong,<sup>1,18</sup> Wolfgang Haak,<sup>1,20</sup> and Johannes Krause<sup>1,20,21,\*</sup>

iScience 25, 104244, May 20, 2022

**19 prehistoric Sicilians covering the Mesolithic to Bronze Age periods (10,700–4,100 yBP).**





# Looking at the origin of fishing in Mediterranean prehistory

NEWS RELEASE 21-FEB-2023

Mediterranean hunter-gatherers relied on marine resources more than previously thought

Peer-Reviewed Publication

UNIVERSITY OF YORK

Mesolithic cemeteries in the Mediterranean, at El Collado, Valencia



**The extent to which humans relied on coastal resources in the past, is key to assessing human health.**

Stable isotope analyses of amino acids reveal the importance of aquatic resources to Mediterranean coastal hunter-gatherers

Maria Fontanals-Coll<sup>1</sup>, Silvia Soncin<sup>1,2</sup>, Helen M. Talbot<sup>1</sup>, Matthew von Tersch<sup>1</sup>, Juan F. Gibaja<sup>3</sup>, André C. Colonese<sup>4</sup> and Oliver E. Craig<sup>1</sup>

Estimation of trophic position using compound-specific isotope analysis of amino acids (CSIA-AA)

PROCEEDINGS B *Proc. R. Soc. B* **290**: 20221330.

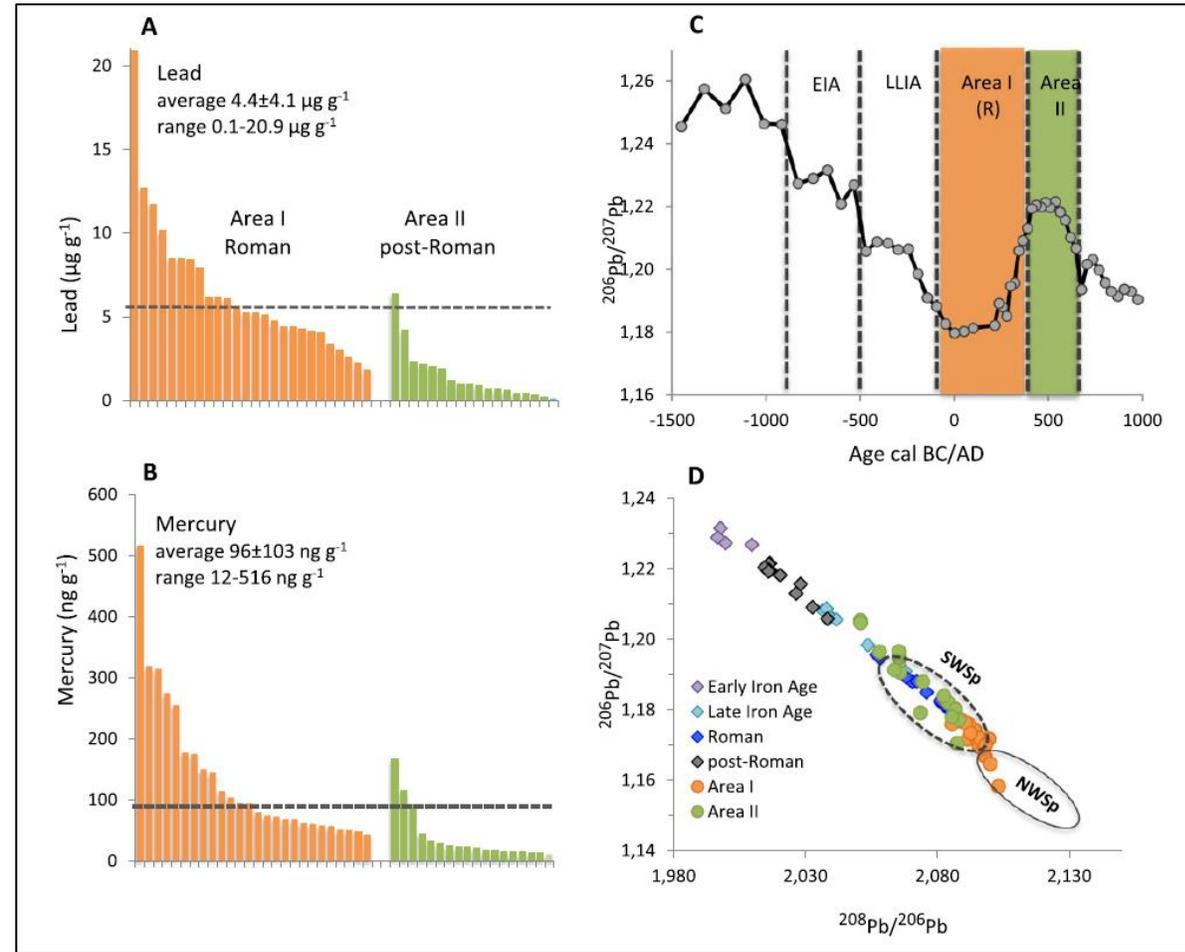
New research has revealed humans living on the Mediterranean coast 9,500 years ago may have relied more heavily on a fish diet than previously thought.

High-resolution biomolecular techniques, like **compound-specific isotope analysis of individual collagen amino acids (CSIA-AA)** allows greater accuracy in discriminating between land animals and marine life.

*López-Costas et al., 2020. Human bones tell the story of atmospheric mercury and lead exposure at the edge of Roman World*



Skeletons are an archive reflecting contaminant exposure.



Lanzada, NWSpain. Roman inhabitants of this settlement incorporated two times more mercury and lead into their bones than post-Romans inhabiting the same site, independent of sex or age.

# Summary



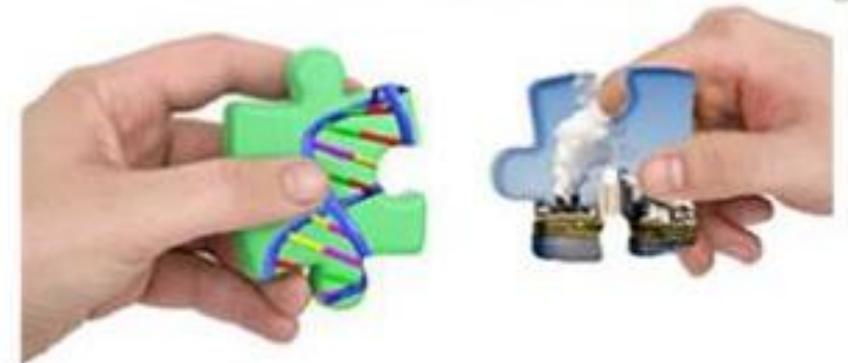
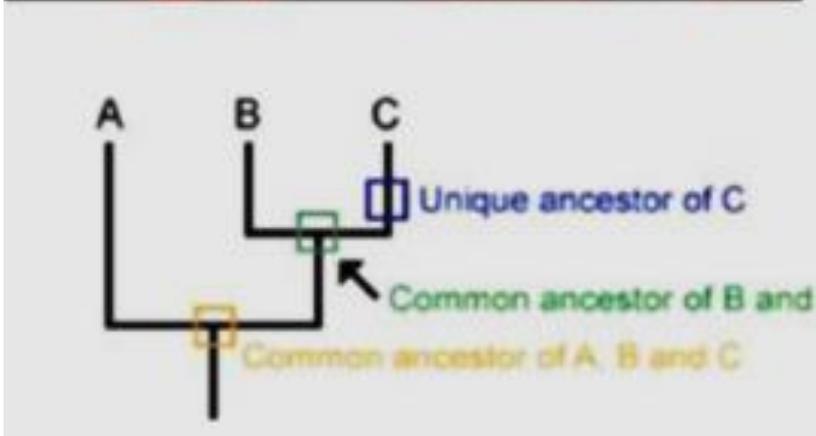
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# **Fish Intake, Contaminants and Human Health**

## **Evaluating the Risks and the Benefits**



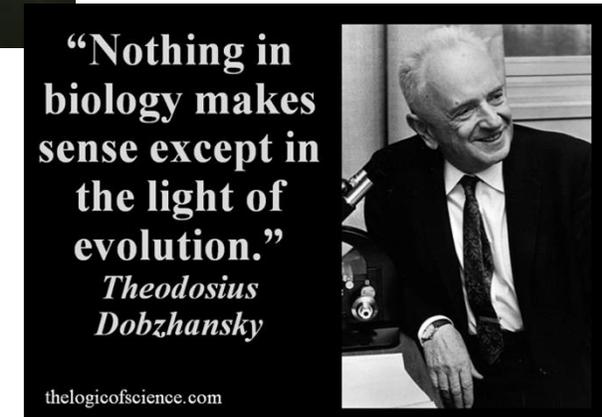
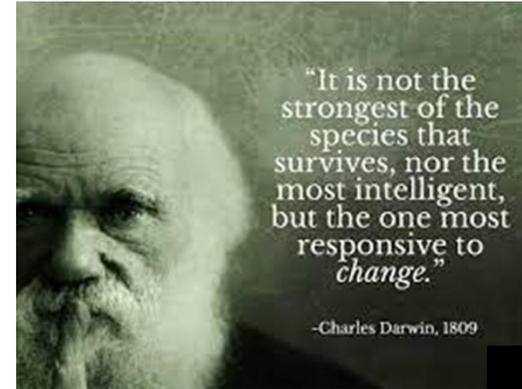
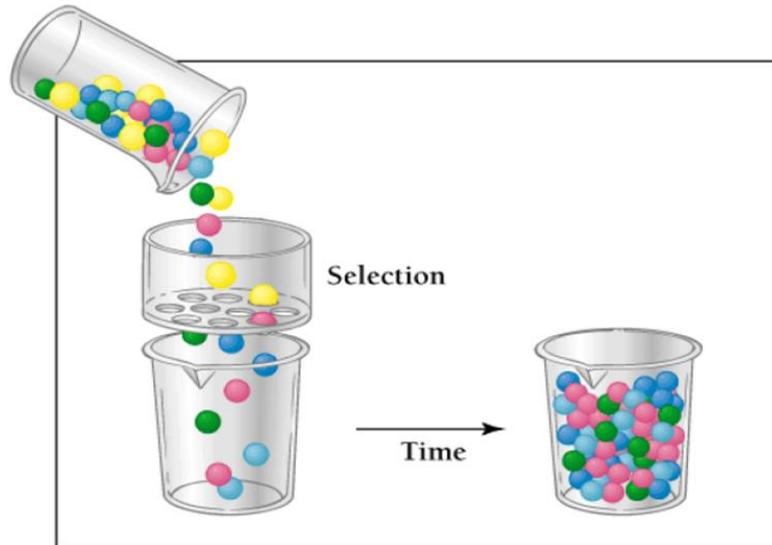
# Human population genomic variability between demography and local adaptation



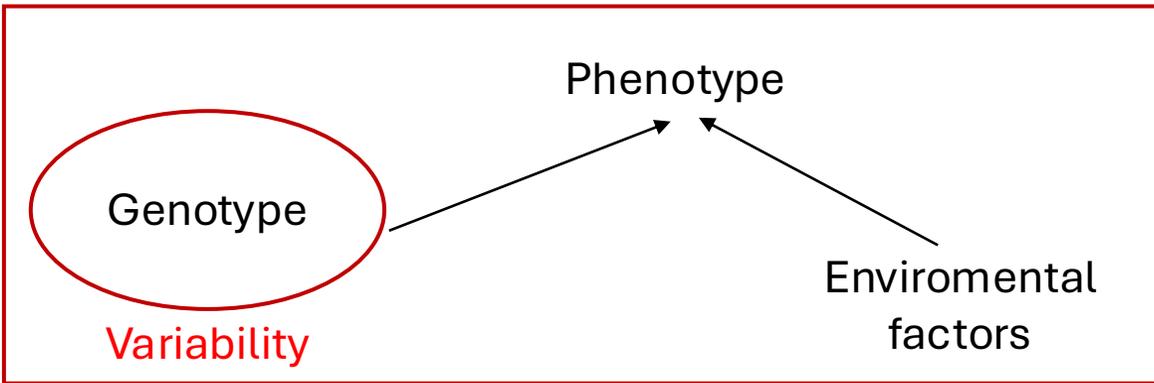
Are marine adaptations true evolutionary adaptations or context-dependent metabolic adjustments?

**Shaping diversity:**

## Selection

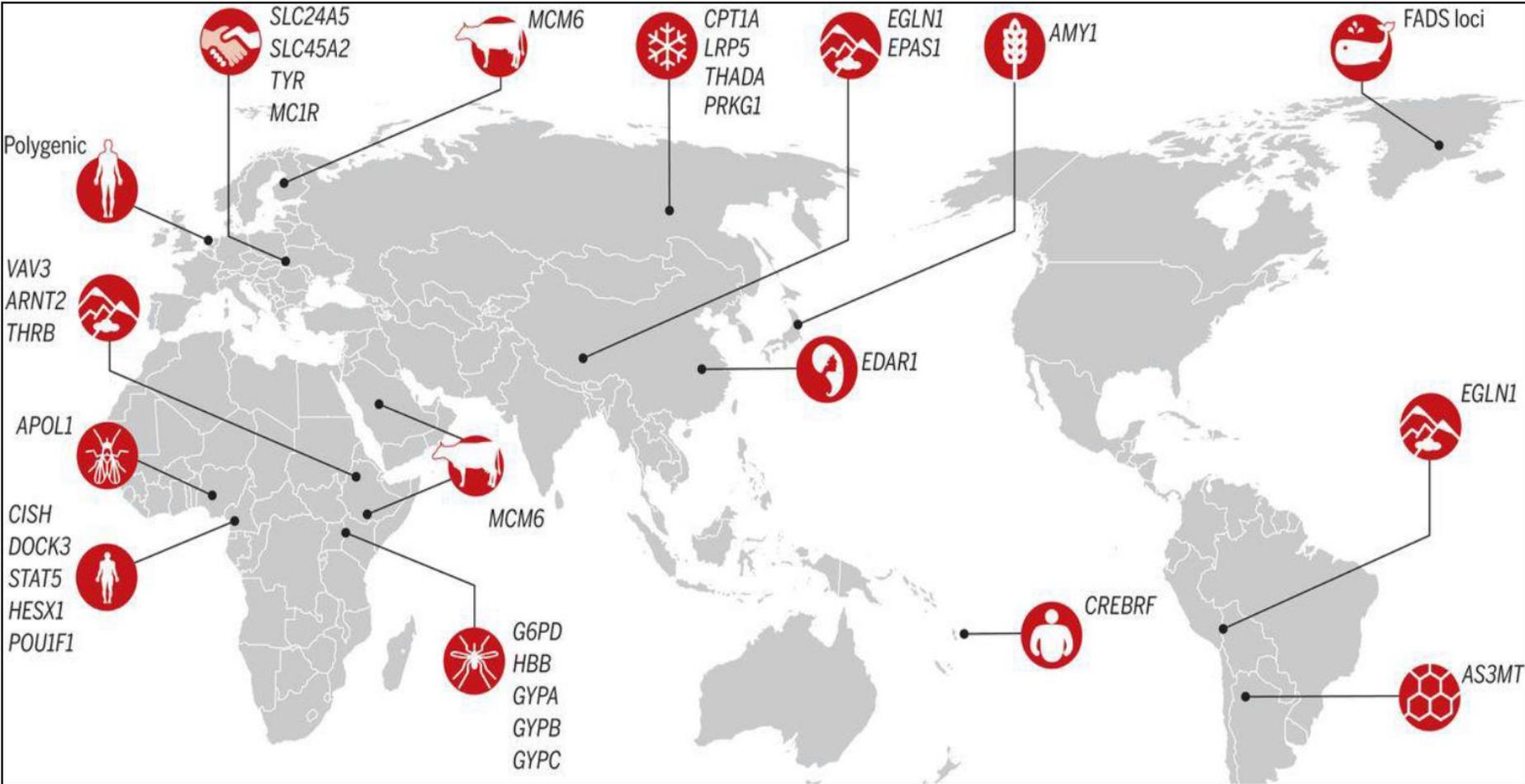


Differential reproduction of genotypes in succeeding generations



*Natural selection outcomes depend at any given moment on the **interaction between environmental and genetic factors.***

- Lactase persistence
- Skin pigmentation
- Height
- High altitude
- Arctic environment
- Trypanosome resistance
- High-fat diet
- Malaria
- Thick hair
- Toxic arsenic-rich environments
- Starchy food
- Increased BMI



# Natural Selection for Human Adaptation in Coastal Environments in modern populations

Humans living in coastal environments have experienced natural selection driven by climate, diet, pathogens, salinity, flooding, and marine-based livelihoods.

Over generations, certain genetic, physiological, and cultural traits increased survival and reproductive success.

## *Genetic Adaptations to Marine-Based Diets*



Coastal populations often rely heavily on seafood rich in omega-3 fatty acids.

# Case Study: Inuit Populations

## Selective Pressure:

- Extremely high intake of **marine lipids (omega-3 fatty acids)**
- Cold climate** increasing metabolic demand

## Genetic Evidence:

Selection signals in genes related to:

- Fatty acid desaturation (**FADS gene cluster**)
- Lipid metabolism regulation
- Cholesterol homeostasis

## Evolutionary Outcome:

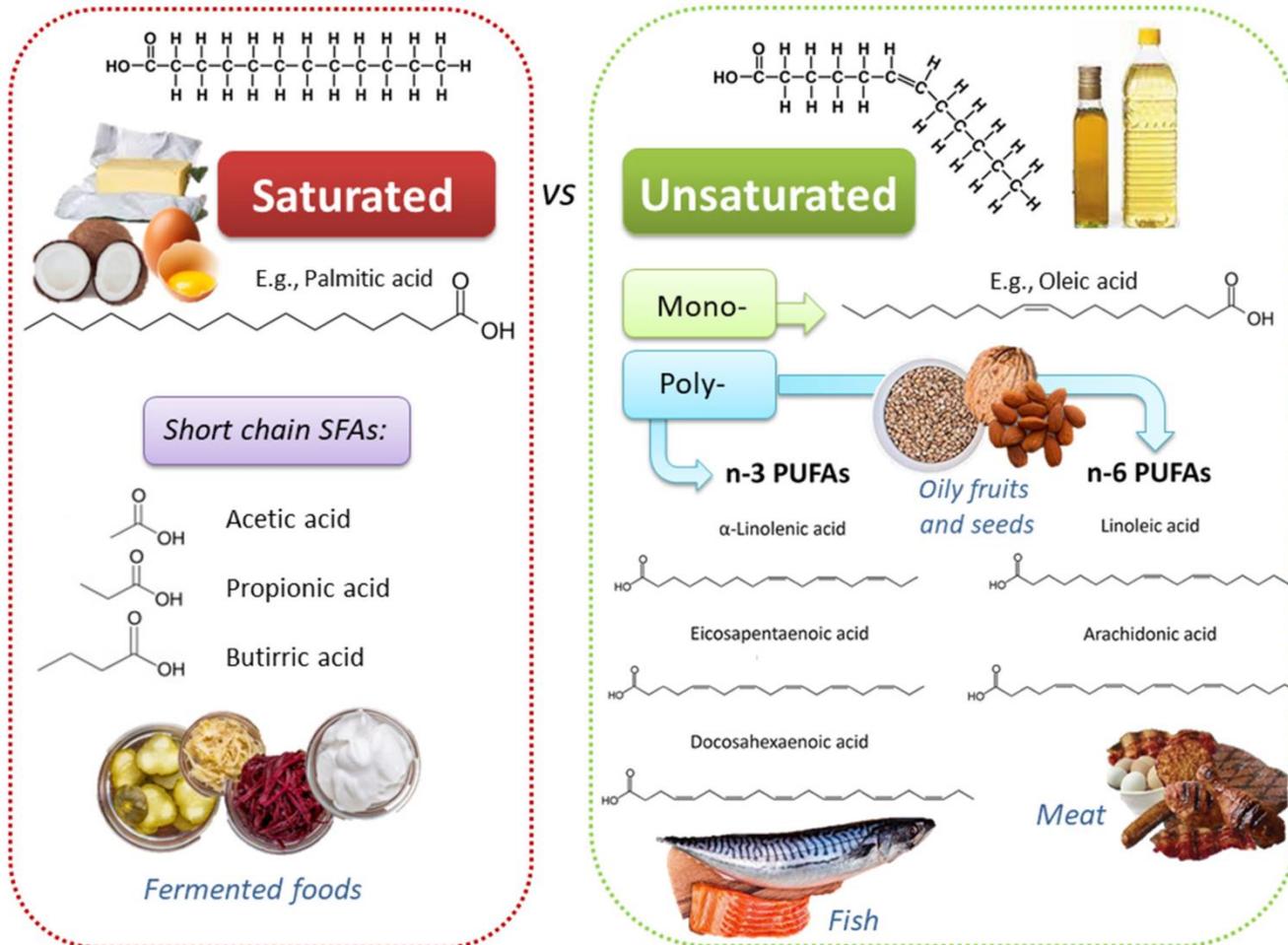
- Modified lipid metabolism
- Lower circulating triglyceride levels
- Cardiovascular regulation suited to marine-fat diet



Review  
**Nutrigenomics of Dietary Lipids**

Laura Bordoni <sup>1</sup>, Irene Petracchi <sup>2</sup>, Fanrui Zhao <sup>2,3</sup>, Weihong Min <sup>3</sup>, Elisa Pierella <sup>4</sup>, Tais Silveira Assmann <sup>5</sup>, J Alfredo Martinez <sup>6,\*</sup> and Rosita Gabbianelli <sup>1,\*</sup>

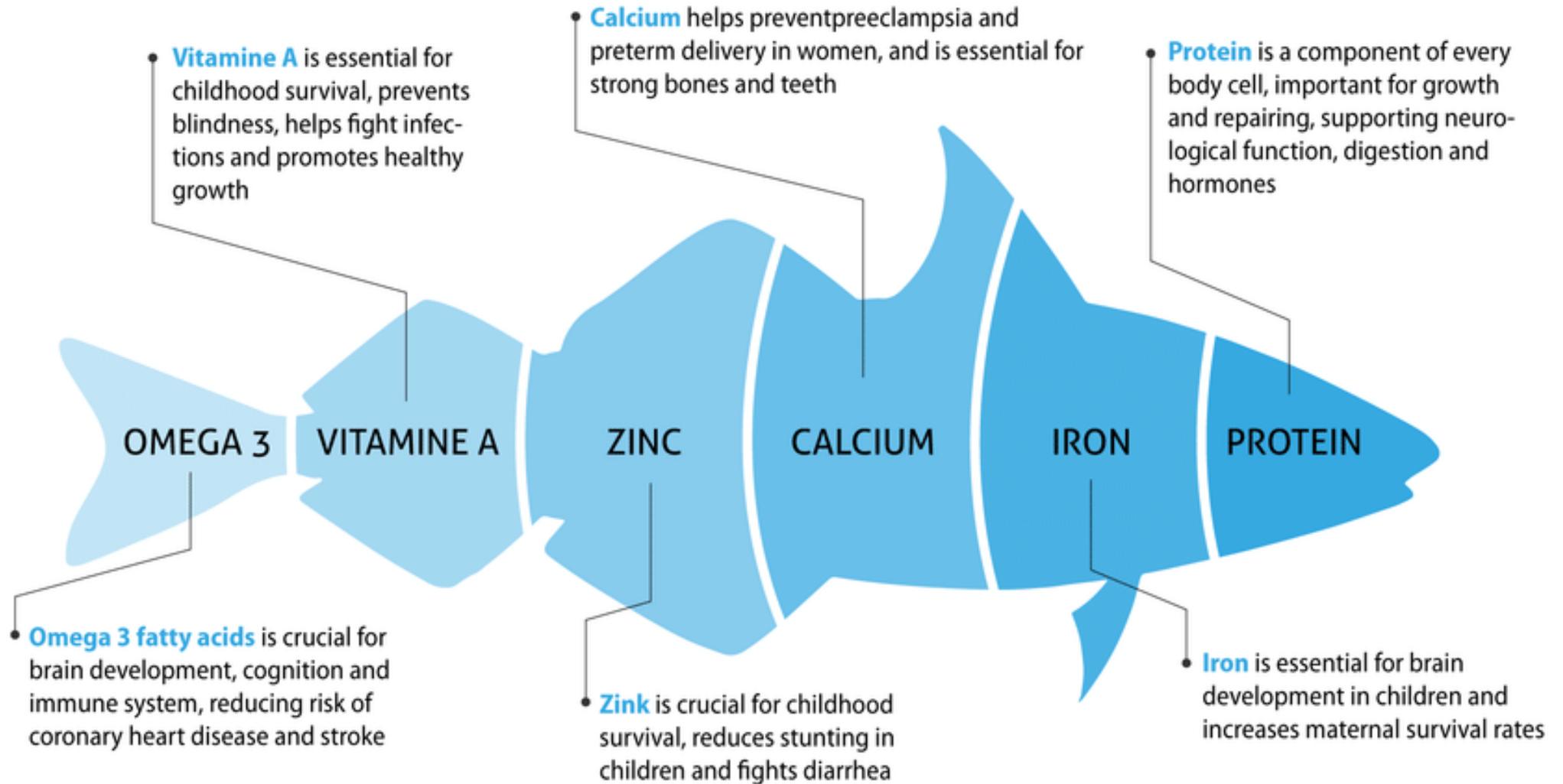
*Antioxidants* **2021**, *10*, 994. <https://doi.org/10.3390/antiox10070994>



Chemically diverse lipid sources, with heterogeneous physico-chemical and biological properties, can be found in different foods.

Food has been studied not only from a chemical prospective, but also for the capacity of metabolites produced by food oxidation to modulate gene expression, directly (nutrigenomics) or by epigenetics remodeling (nutriepigenomics).

However, **inter-individual differences (i.e., the genetic variability) and environmental exposures** (i.e., physical activity, drug, food pesticide residues, etc.) contribute to produce a lot of effects

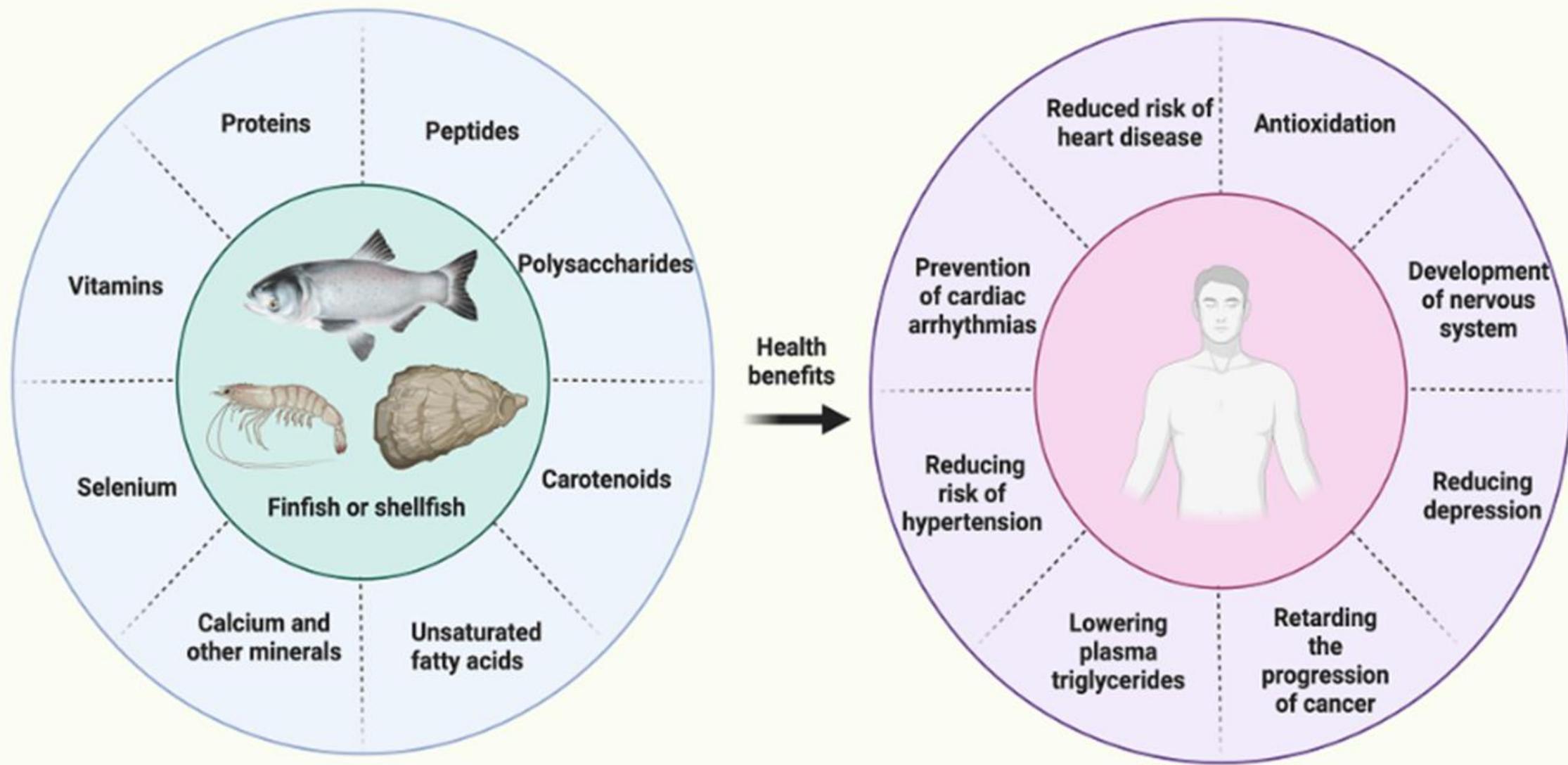


 **Toxics:** some seafood can accumulate heavy metals, dioxine, PCB, ciguatoxin and antibiotic residuals.

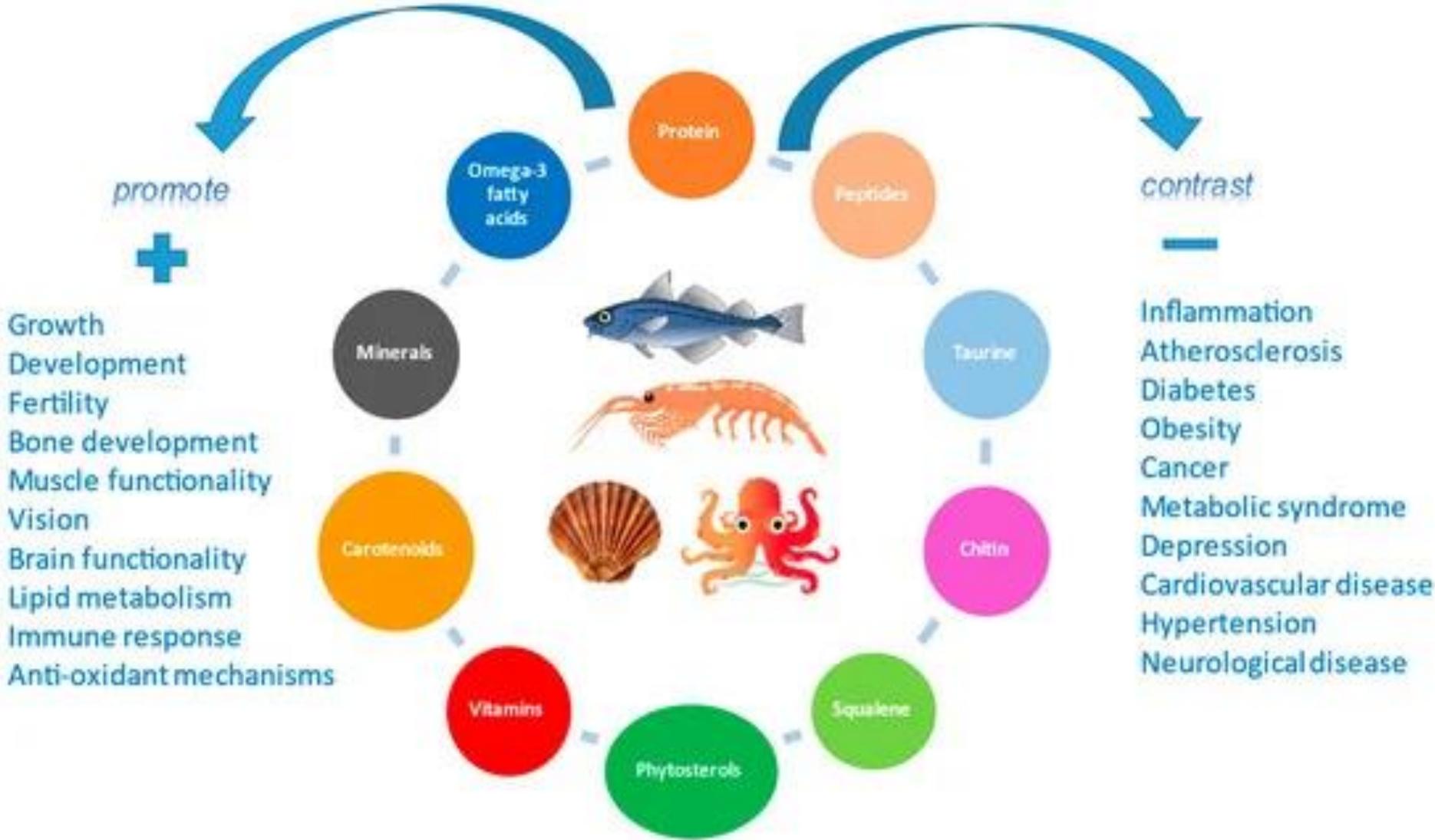
# Special Issue "Fish Intake and Human Health: Evaluating the Nutrients and Benefits"

- **Fish (finfish or shellfish) are essential to a healthy diet.**
- Fish are the primary sources of healthy long-chain omega-3 fats and are rich in other nutrients such as vitamin D and selenium, high in protein, and low in saturated fat.
- There is strong evidence that **eating fish or taking fish oil has a positive impact on the heart and blood vessels.**

## Fish Intake and Human Health: Evaluating the Nutrients and Benefits



# HEALTH EFFECTS



# Nutritional Ecology of Traditional Fishing Communities

## Macronutrient Structure

Traditional marine diets typically exhibit:

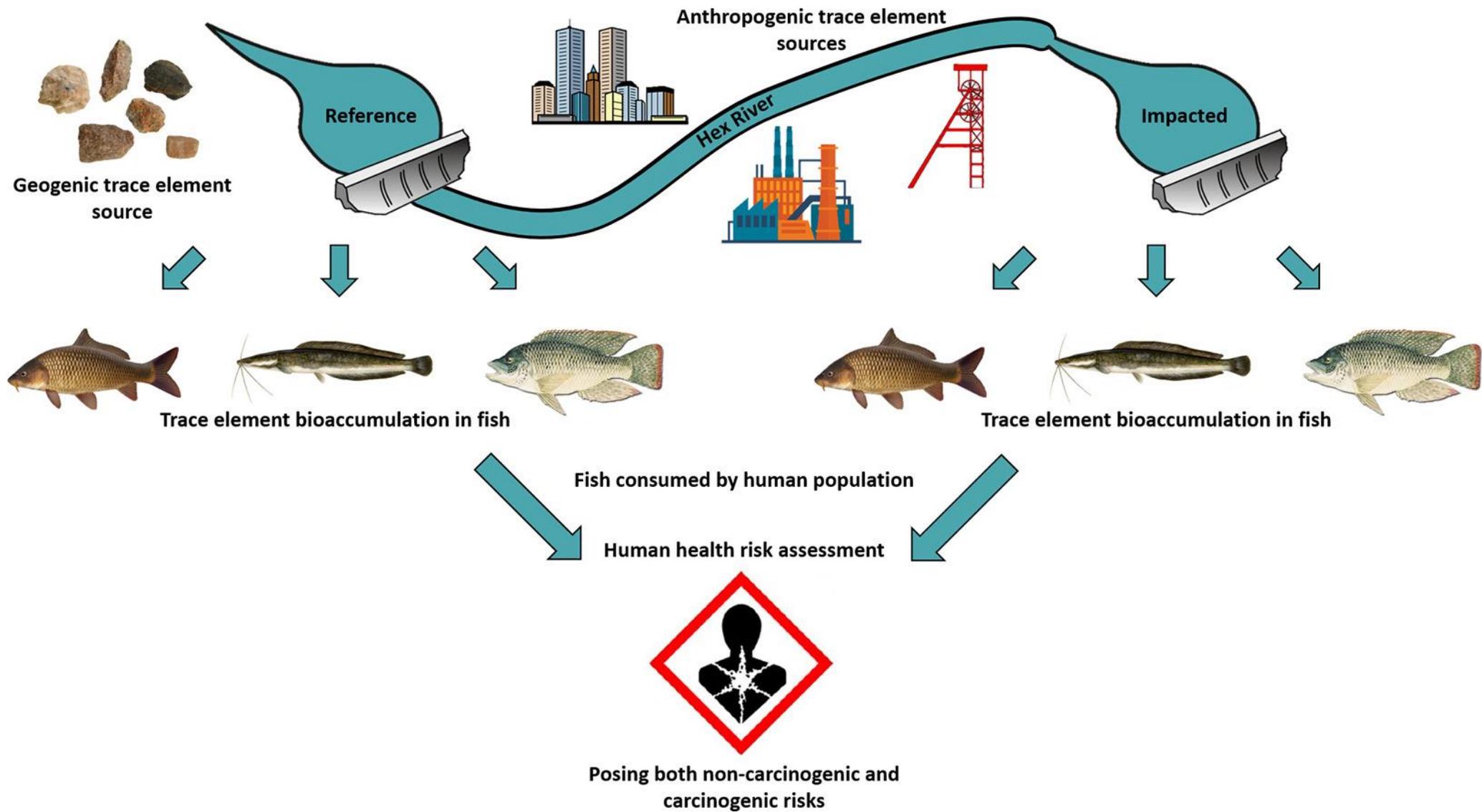
- High protein intake (25–30% energy)
- High omega-3 fatty acid intake
- Low refined carbohydrate intake

Such dietary structures modulate lipid metabolism and inflammatory signaling pathways (Calder, 2023).

## Micronutrients:

Marine ecosystems provide **high iodine availability**, supporting thyroid function. Historically, inland iodine deficiency led to endemic goiter, whereas coastal fishing populations showed lower prevalence. Thyroid regulation plays a central role in metabolic homeostasis.

**Selenium intake may also mitigate mercury toxicity**, introducing **a complex risk–benefit balance.**



Among the main seafood contaminants there are organochlorine pesticides, organotin compounds, phthalates, brominated flame retardants, polyfluorinated compounds, polycyclic aromatic hydrocarbons (PAH), dioxins, dioxin-like PCBs and non-dioxin-like PCBs, heavy metals (mercury, cadmium, lead) and arsenic

# Environmental Toxicology

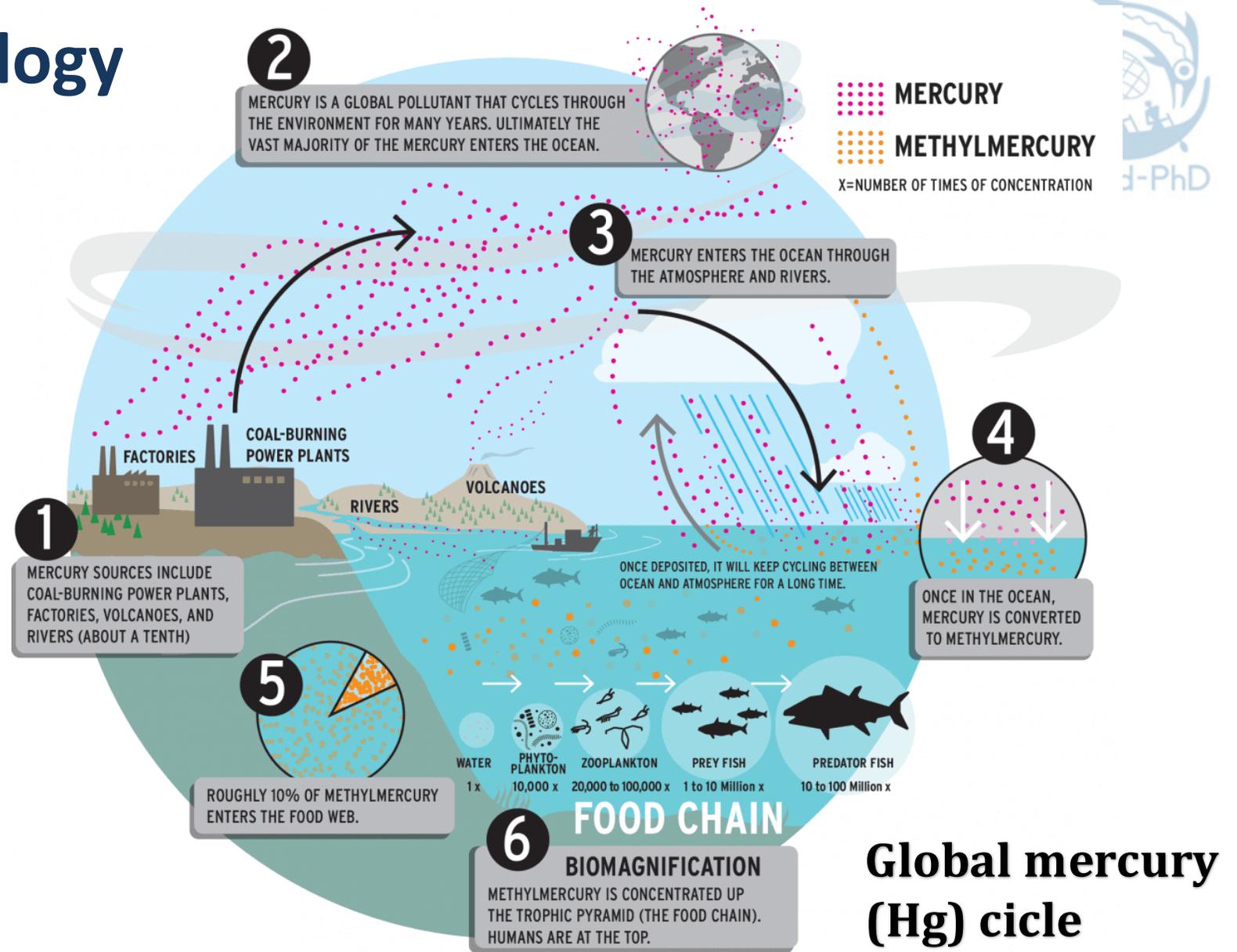
## Mercury

## Bioaccumulation

Mercury concentration increases with trophic level due to biomagnification. Long-lived predatory fish (e.g., tuna, seal) contain high methylmercury levels.

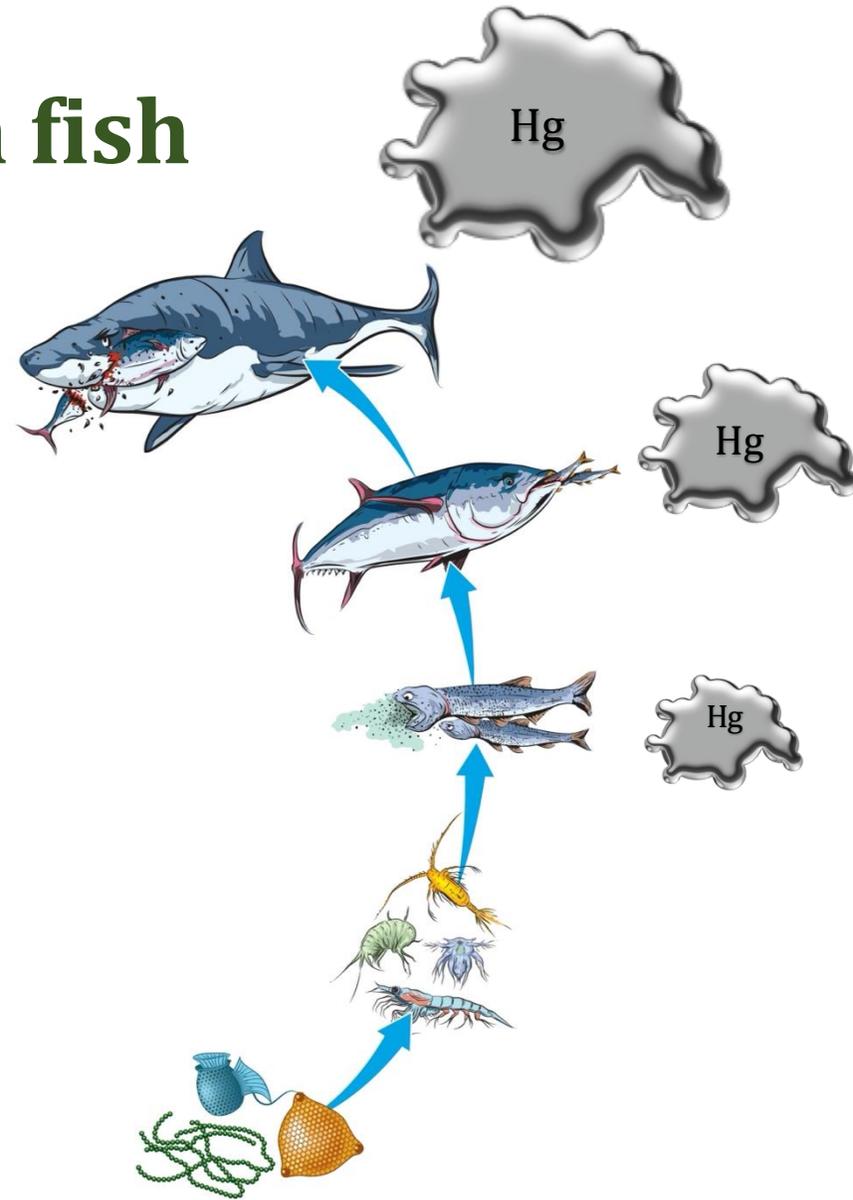
### Health impacts include:

- Neurodevelopmental impairment
- Cardiovascular risk
- Epigenetic modifications (FAO/WHO, 2023)



# Mercury concentration in fish increases with:

- Trophic level
- Size
- Age
- Demersal habitat



# *Persistent Organic Pollutants (POPs)*

Arctic populations experience **high exposure** due to atmospheric transport and fat-rich marine diets.

POPs disrupt endocrine signaling and may contribute to metabolic dysregulation.

Unlike the Arctic, the Mediterranean is semi-enclosed basin with high coastal population density and heavy maritime traffic. Contamination is regionally produced and regionally retained.

While **Arctic fishing populations** represent a model of extreme biomagnification and long-term contaminant exposure potentially shaping detoxification pathways, **Mediterranean fishing populations** illustrate regionally driven, industrial-era pollutant exposure superimposed on a historically mixed subsistence system.

Detoxification in fishing populations is not merely a toxicological issue, but an interface between ecological exposure, metabolic adaptation, and **human genetic polymorphism**.

### **Risk–Benefit Dynamics**

#### **Arctic**

High benefit (omega-3, vitamin D, iodine); high risk (POPs, mercury); clear evolutionary trade off system.

#### **Mediterranean**

Moderate benefit (fish consumption; moderate but spatially variable risk, diet is more diverse → toxic load is diluted.

**High omega-3 intake (Arctic)** may modulate inflammatory signaling; buffer oxidative damage and alter expression of detox genes

**Mediterranean mixed diet** may provide antioxidants (polyphenols); reduce oxidative burden and modify toxicokinetics

**Diet–gene–pollutant interaction is central.**

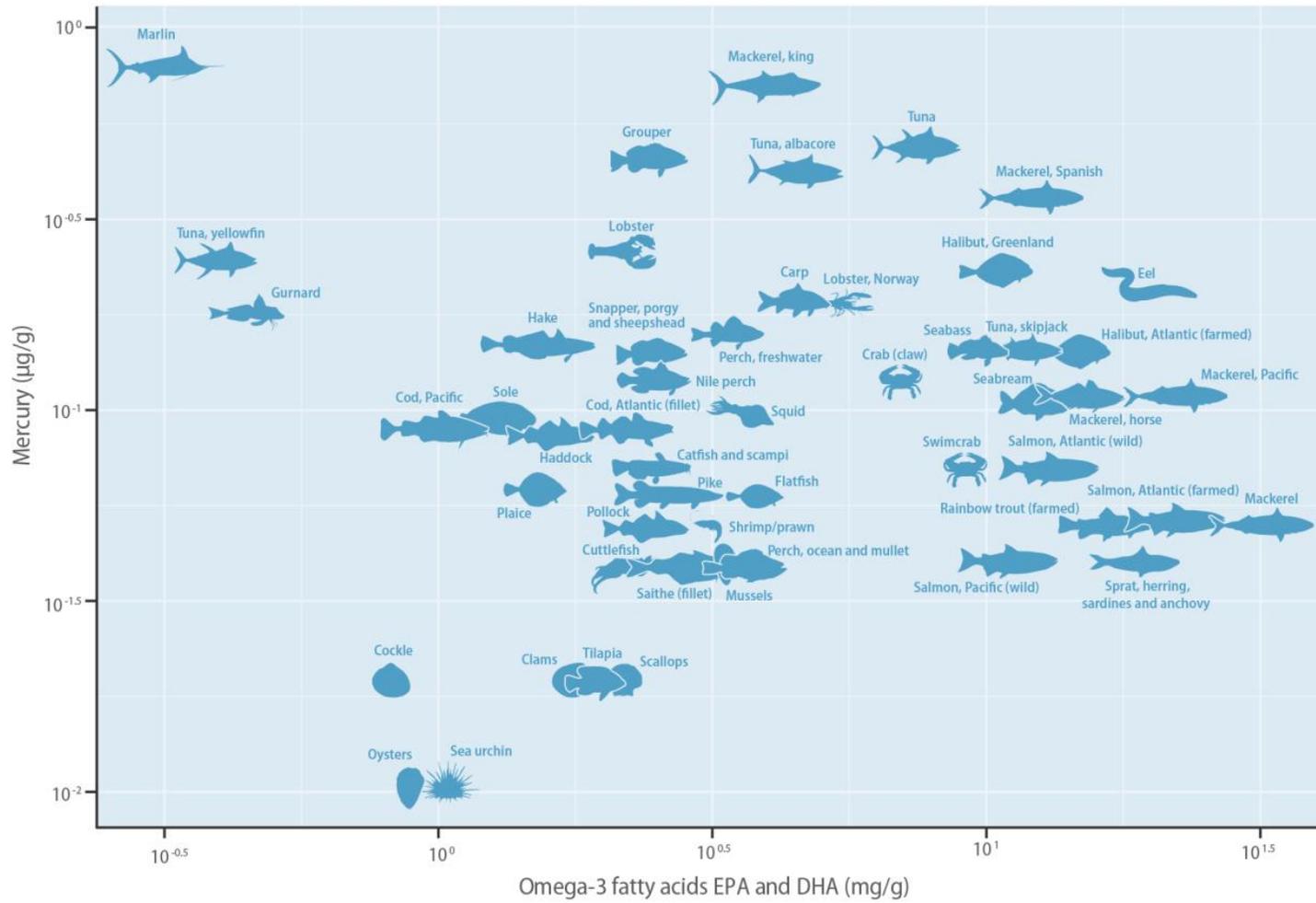


Fig 7. Illustration of omega-3 fatty acids and Mercury (Hg) in various seafood (From <sup>68</sup>)

Figure 2: Genetic Adaptations to Marine Diets

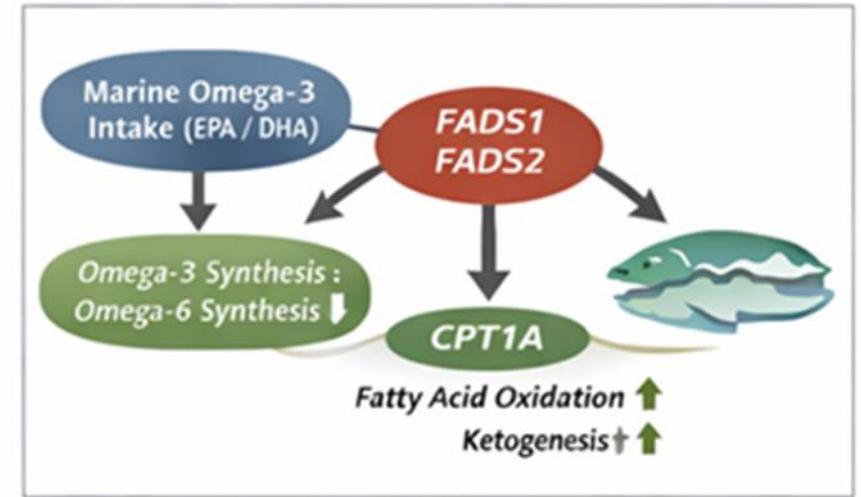
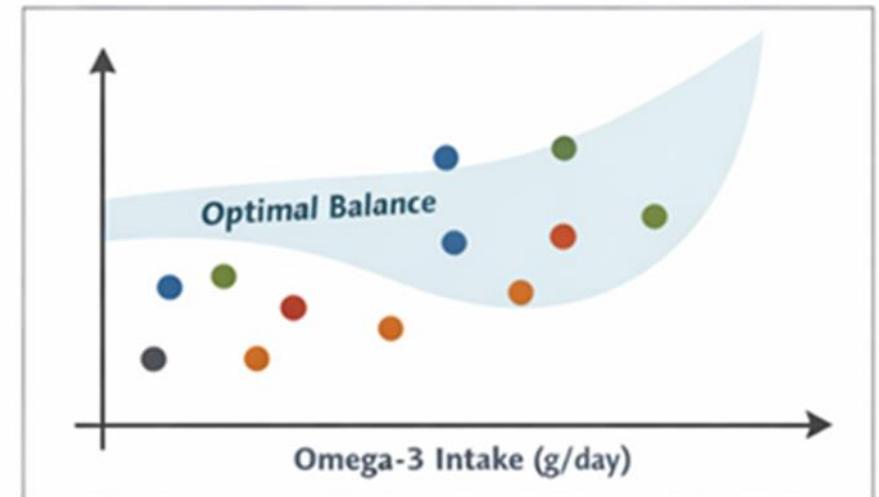


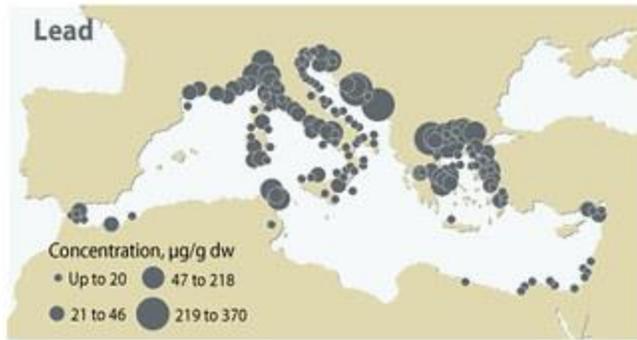
Figure 4: Omega-3 Intake vs. Contaminant Exposure



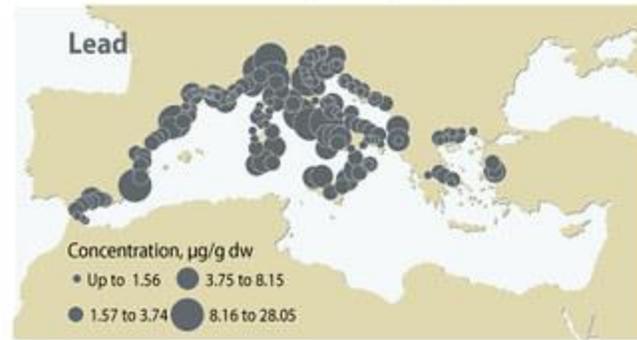


## Mean concentrations of trace metals

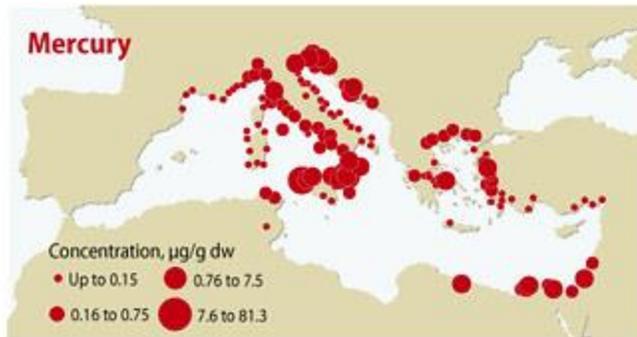
### In sediments



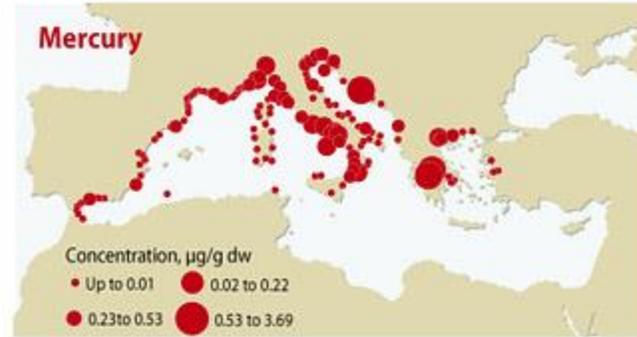
### In Blue Mussels (*Mytilus galloprovincialis*)



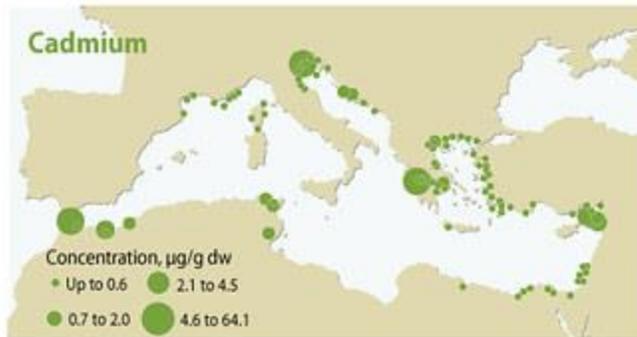
### Mercury



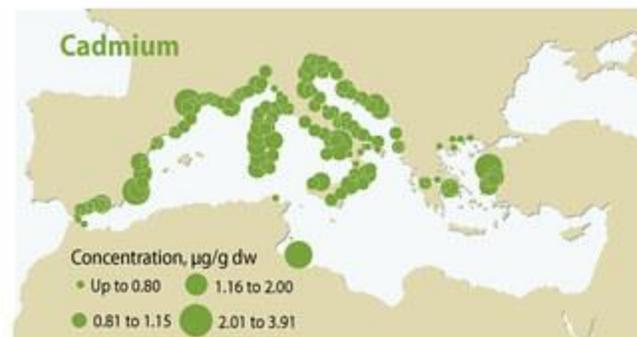
### Mercury



### Cadmium

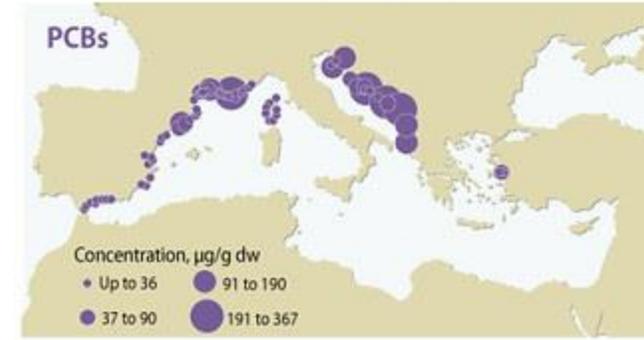


### Cadmium



## Mean concentrations of Persistent Organic Pollutants (POPs)

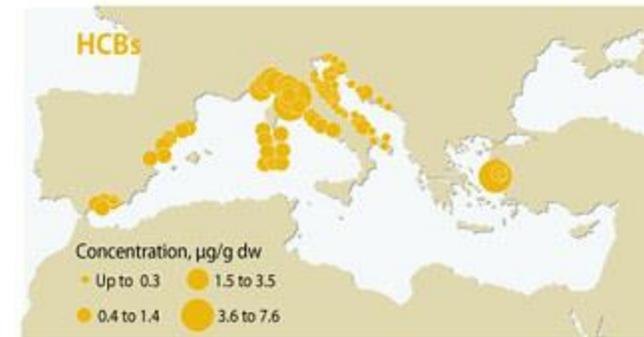
### In Blue Mussels (*Mytilus galloprovincialis*)



### DDTs



### HCBs



Source: UNEP/MAP, Hazardous Substances in the Mediterranean: A Spatial and Temporal Assessment, 2011

## Traditional fishing communities historically displayed:

- Low atherosclerosis
- High infectious disease burden
- Seasonal nutritional stress

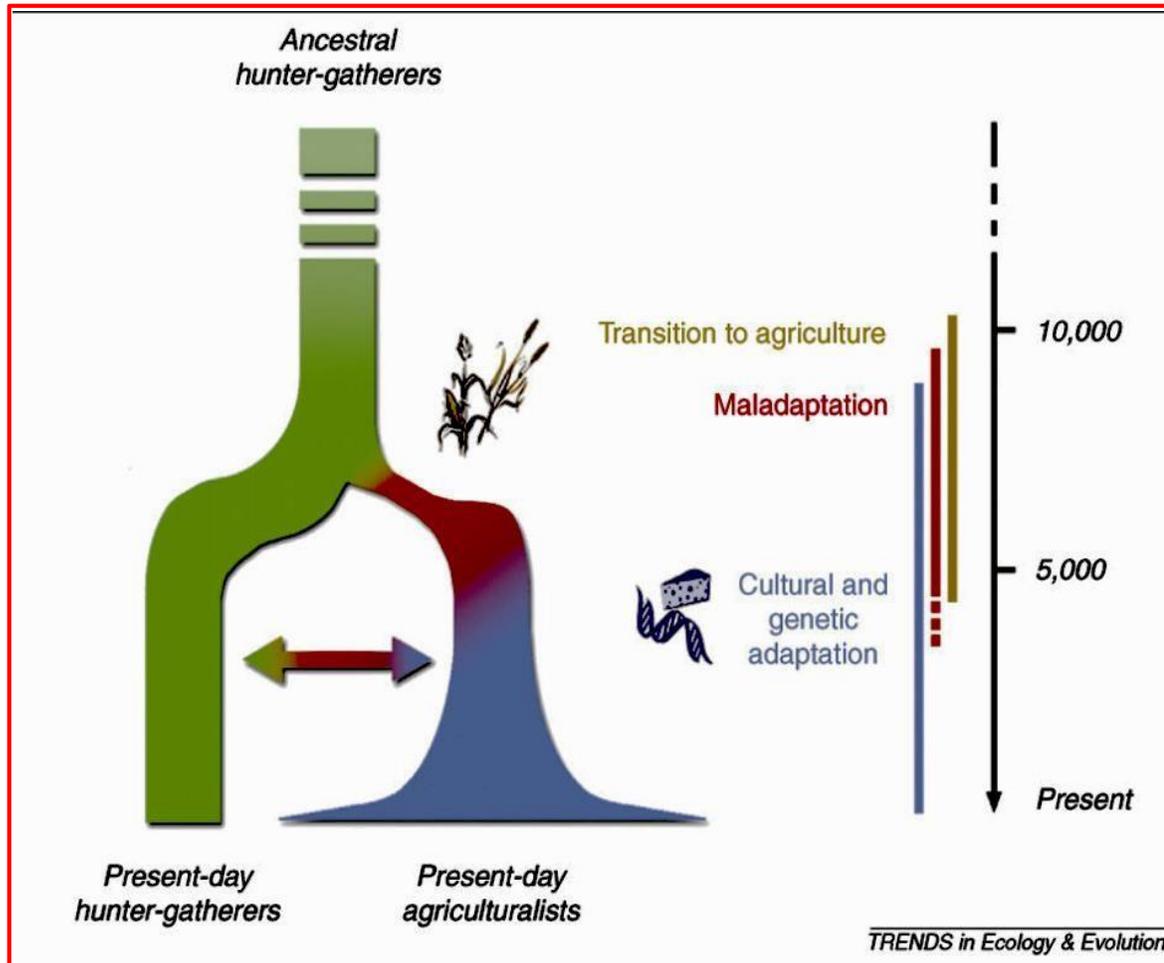
## Modern transition introduces:

- Refined carbohydrates
- Ultra-processed foods
- Reduced physical activity

Rapid metabolic syndrome emergence in Arctic and Pacific Island populations exemplifies evolutionary mismatch: genomes adapted to specific ecological conditions now operate within radically altered nutritional environments.

The **mismatch hypothesis** proposes that genomes adapted to specific nutritional ecologies become maladaptive under modern dietary conditions (Cordain et al., 2005).

# Trade-offs past selection : **maladaptation**

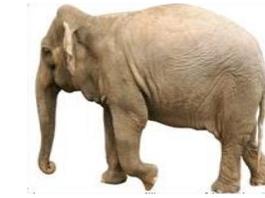


## MISMATCH

The poorly adapted phenotype in Western countries resulting from the conflict between the changing lifestyle and our Paleolithic genome.



PALEOLITHIC GENOME



FAST CHANGING OF LIFESTYLE



PATHOLOGY OF WESTERN COUNTRIES

# Summary



- Traditional fishing communities as evolutionary “natural laboratories”
- Marine resources from past to present
- Fishing: health benefits and contaminants
- **Genetics and epigenetics of fishing communities**

# The FADS Gene Cluster

The FADS1 and FADS2 genes encode enzymes responsible for endogenous synthesis of long-chain polyunsaturated fatty acids (LC-PUFAs). In populations consuming high levels of preformed marine omega-3 fatty acids (EPA, DHA), selection appears to favor variants that reduce endogenous synthesis.

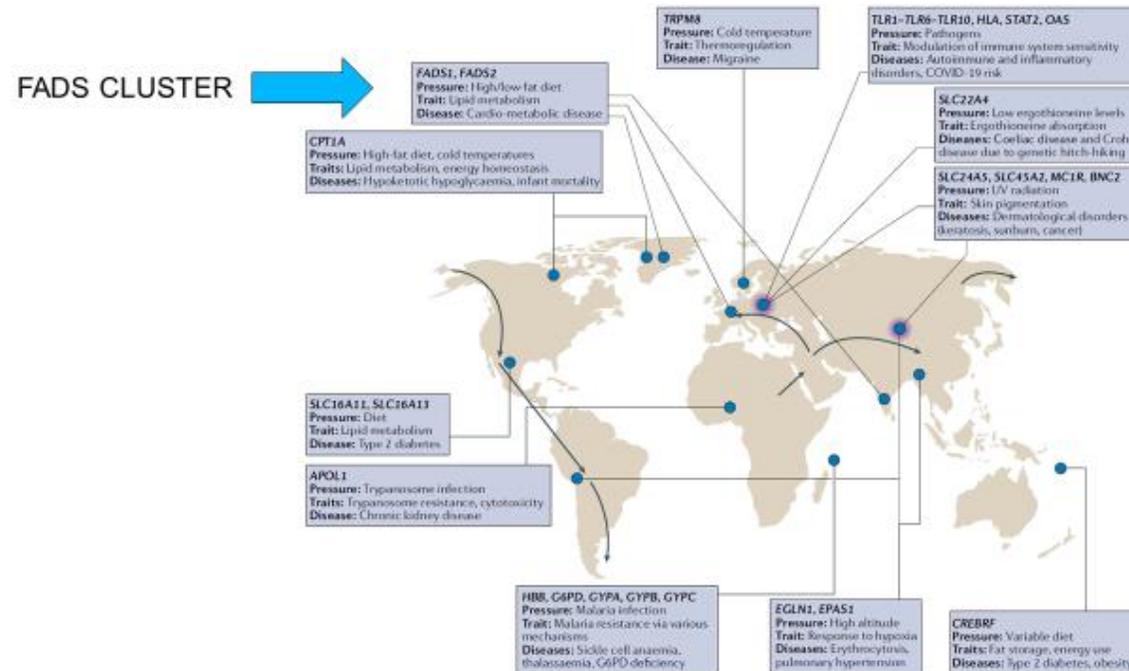


Fig. 2 | Recent adaptation has produced evolutionary trade-offs that lead to disease in some environments. Representative genes that have experienced local adaptive evolution over the past 100,000 years as humans moved across the globe. We focus on adaptations that also produced the potential for disease due to trade-offs or mismatches with modern environments. For each, we list the evolutionary pressure, the trait(s)

influenced and the associated disease(s). The approximate regions where the adaptations occurred are indicated by blue circles. Arrows represent the expansion of human populations, and purple shading represents introgression events with archaic hominins. Supplementary Table S1 presents more details and references. COVID-19, coronavirus disease 2019; G6PD, glucose-6-phosphate dehydrogenase; UV, ultraviolet.



FishMed-PhD

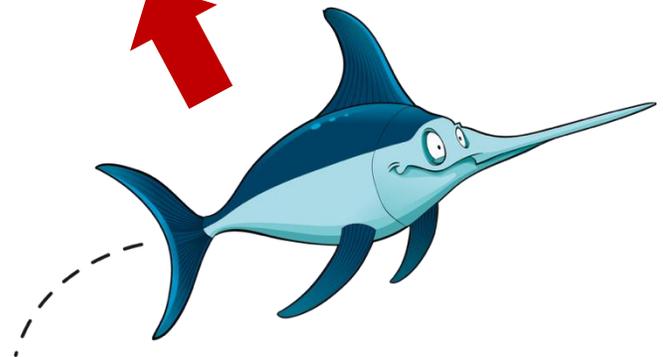
***FADS1***  
***FADS2***



delta-5 and delta-6  
desaturases



eicosapentaenoic acid (EPA)  
docosapentaenoic acid (DHA)

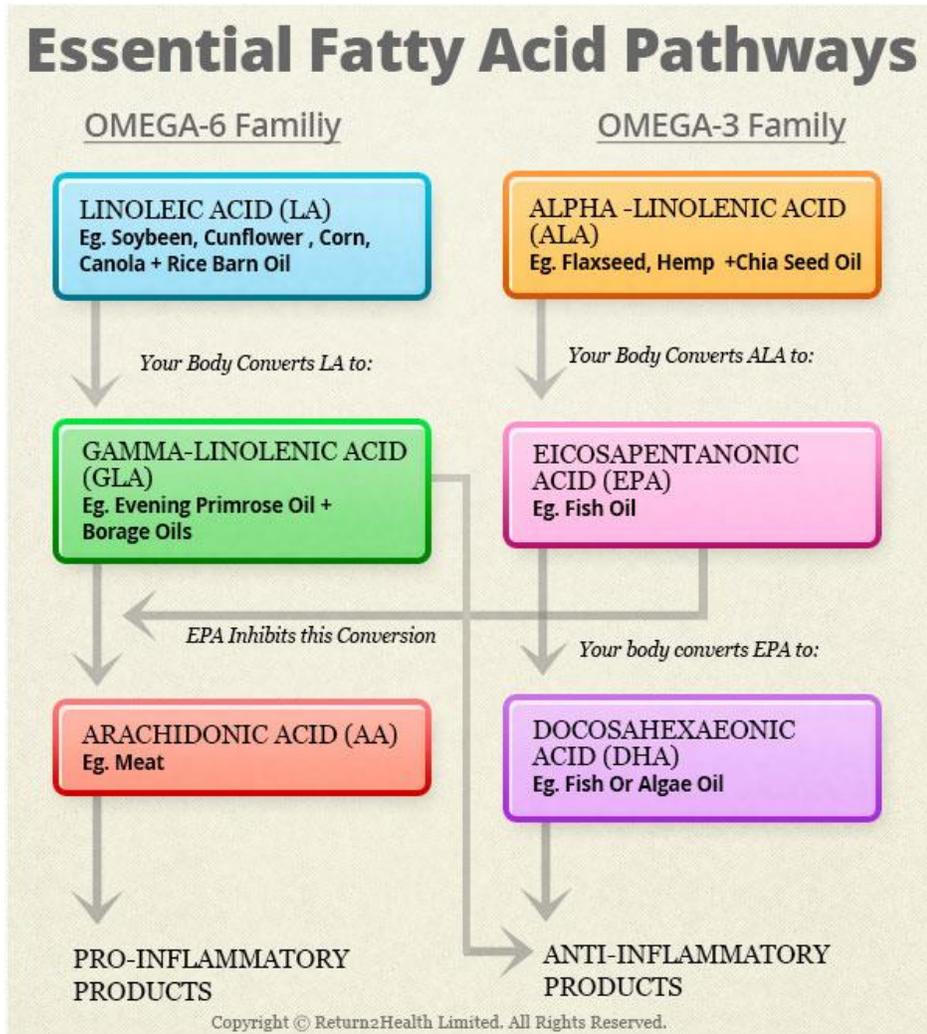


linoleic acid (LA)  
 $\alpha$ -linolenic acid (ALA)



# EVOLUTIONARY ASPECTS OF THE DIETARY OMEGA6/OMEGA 3 FATTY ACID RATIO

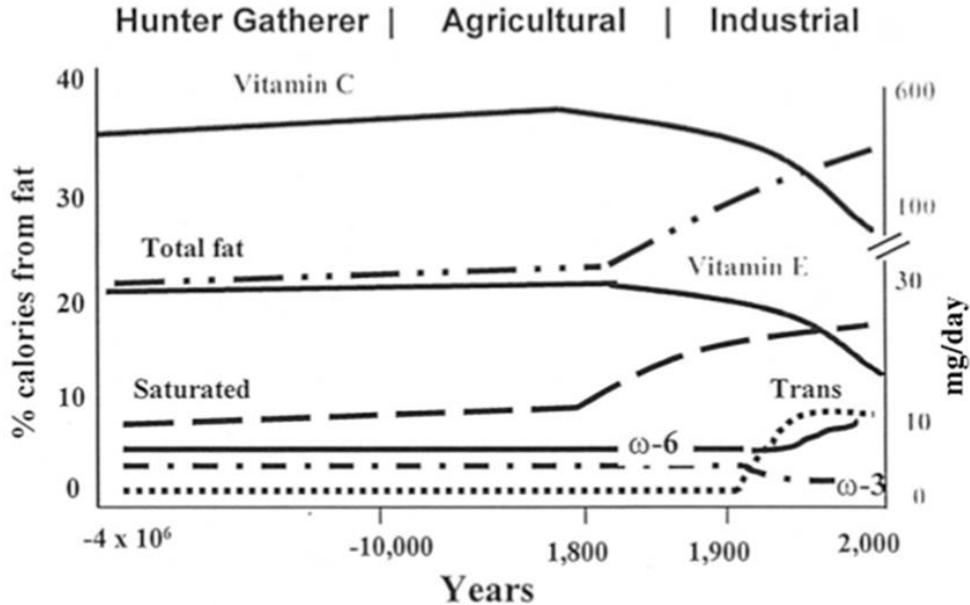
- a balance existed during the long evolutionary history of genus *Homo*;
- **essential to humans in the sense that they cannot be synthesized de novo but need to be supplied through **dietary intake**;**
- lack of the converting enzyme, omega-3 desaturase (omega-6 to omega-3 fatty acids);
- a **balance** is a physiological state (important physiological effects)



The metabolic conversion of dietary omega-3 and omega-6 18 carbon (18C) to long chain (>20 carbon) polyunsaturated fatty acids (LC-PUFAs) is vital for human life.

The rate-limiting steps of this process are catalyzed by fatty acid desaturase FADS 1 and 2

During the Paleolithic period, the diets of humans included equal amounts of omega-6 and omega-3 fatty acids from plants (LA+ ALA) and from the fat of animals in the wild and fish (AA + EPA + DHA).

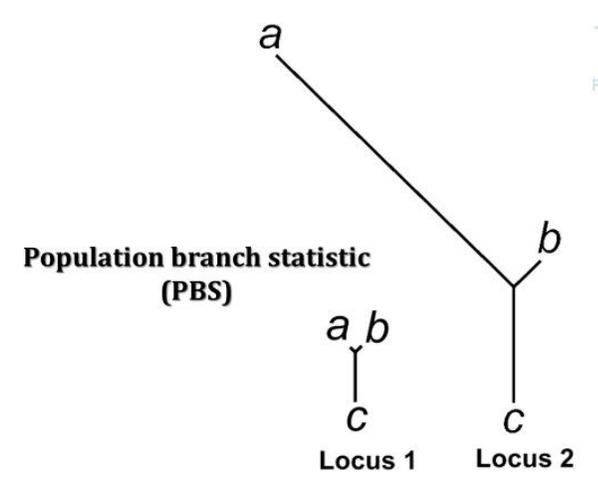
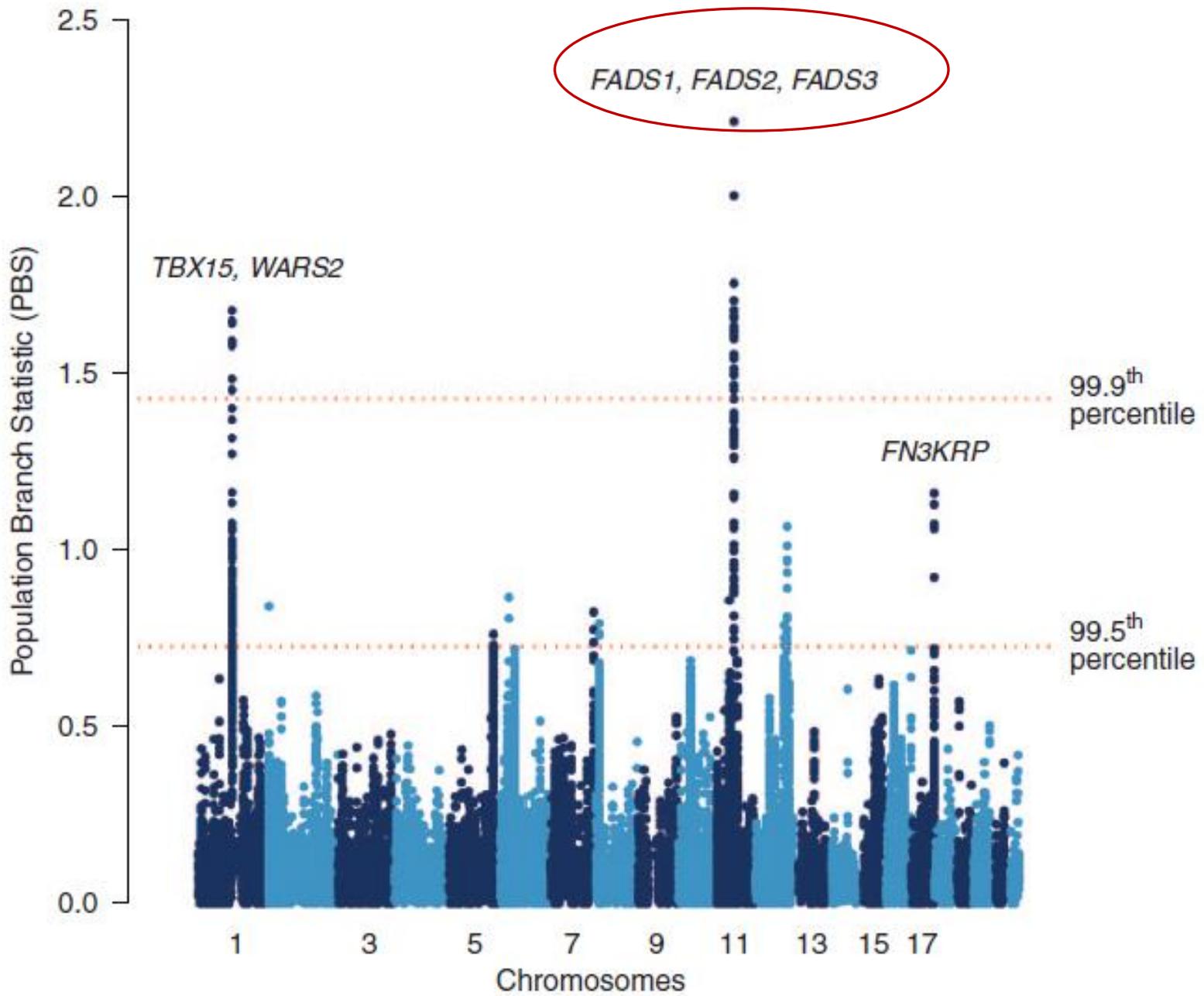


Rapid dietary changes over short periods of time have occurred **over the past 100–150 years** → totally new phenomenon in human evolution.

Population	$\omega$ -6/ $\omega$ -3
Paleolithic	0.79
Greece prior to 1960	1.00–2.00
→ Current Japan	4.00
Current India, rural	5–6.1 ←
Current UK and northern Europe	15.00
Current US	16.74
Current India, urban	38–50 ←

**Urbanization** and **industrialization** processes change the ratio

*Omega-6/Omega-3 ratios in different populations*



Fumagalli et al., 2015. Greenlandic Inuit show genetic signatures of diet and climate adaptation

A scan of Inuit genomes for signatures of adaptation revealed signals at several loci, with the strongest signal located in a cluster of fatty acid desaturases that determine PUFA levels.

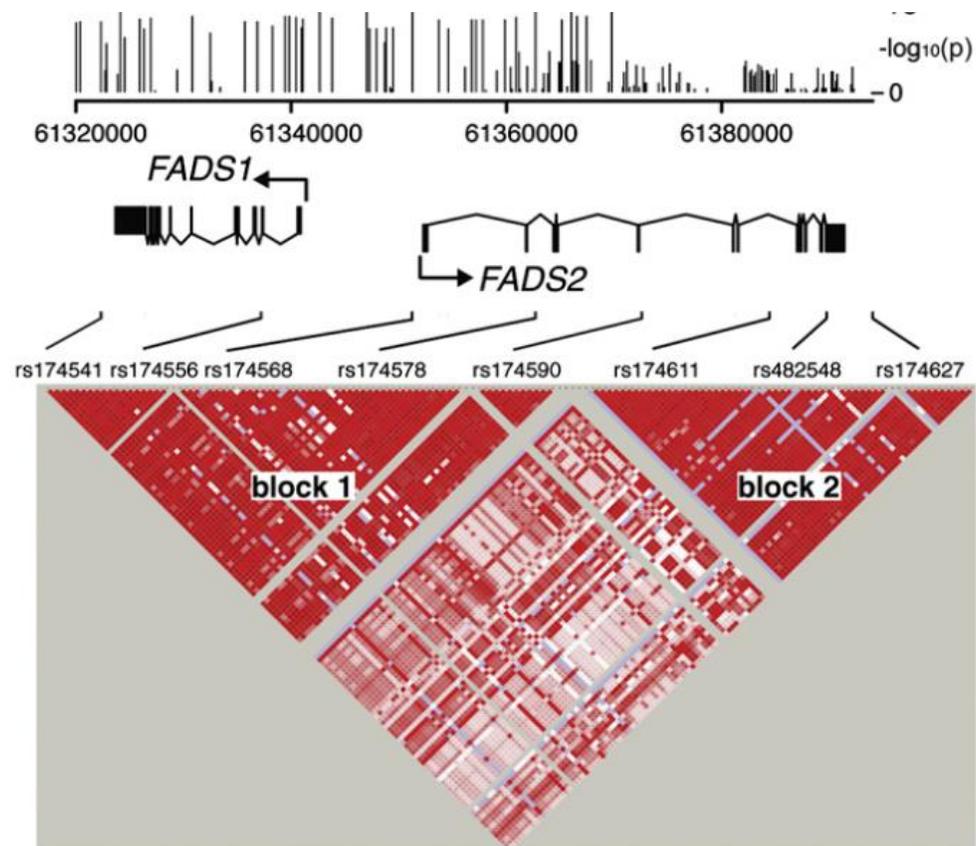
LD Block 1

LD Block 2

*FADS1*

*FADS2*

*FADS3*



## LD and HAPLOTYPIC BLOCK

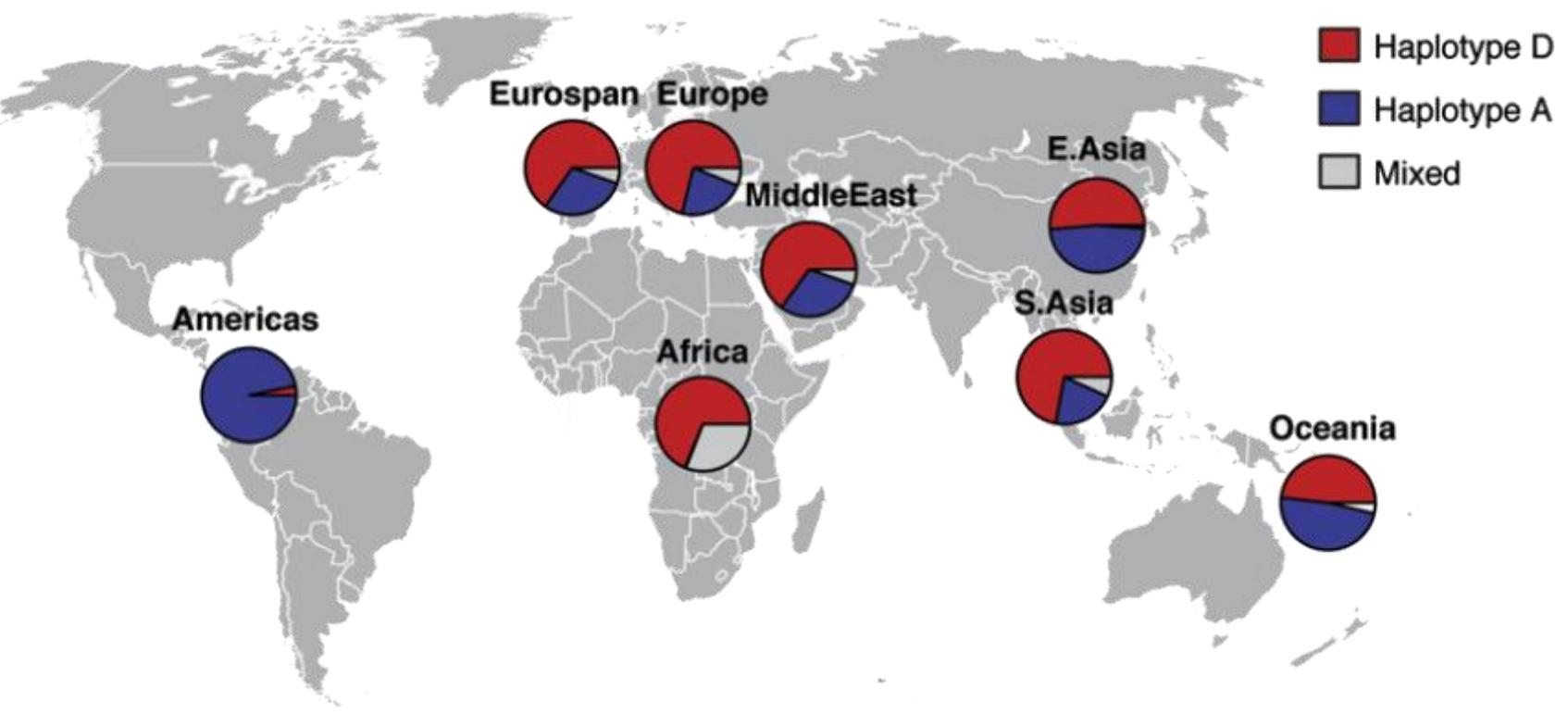
Selection has targeted **different alleles in the FADS genes in Europe than it has in South Asia or Greenland.**

**LD BLOCK 1: FADS1 + 1° half of FADS2**

**HAPLOTYPE A**

**HAPLOTYPE D**

(increased FADS1 activity and is hypothesized to be an adaptation to a diet relatively low in PUFA, whereas haplotype A is hypothesized to be advantageous in a PUFA-rich environment).



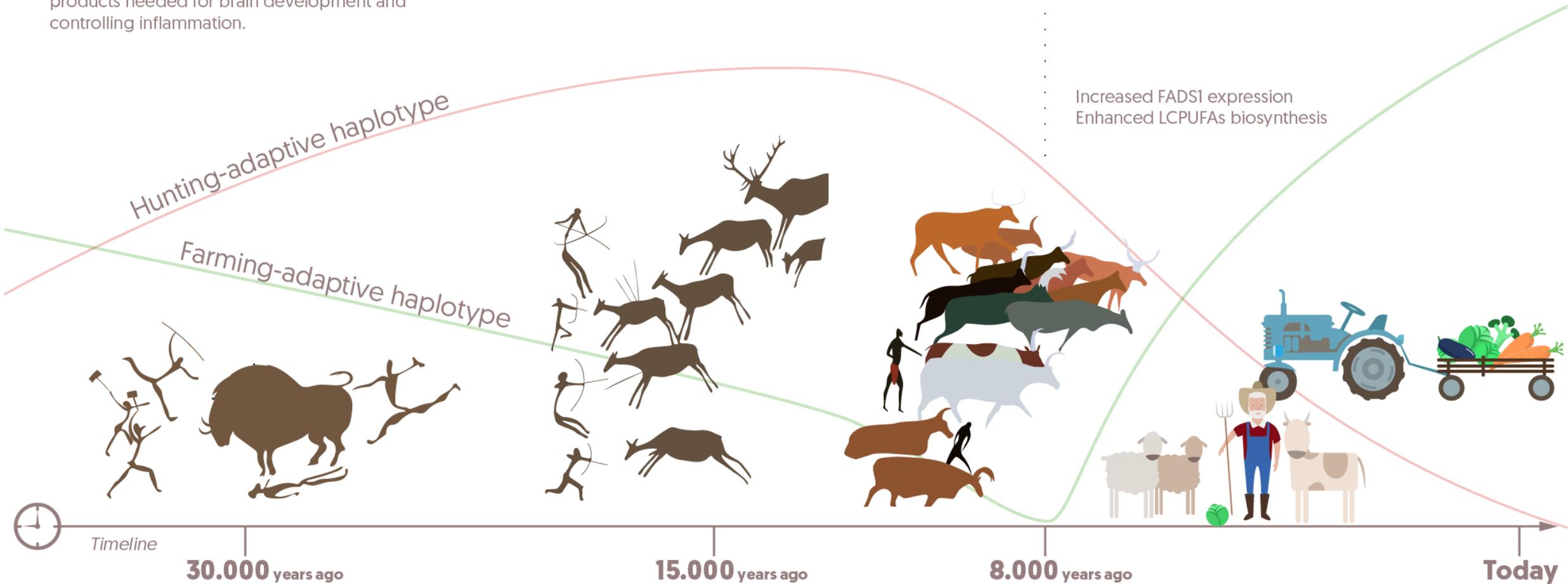
*Haplotype* - group of genes at multiple locations in a single chromosome.

FADS1 and FADS2 are enzymes that are essential to metabolize plants and for converting omega-3 and omega-6 fatty acids (LCPUFA) into downstream products needed for brain development and controlling inflammation.



## The Neolithic Revolution in Europe

Increased FADS1 expression  
Enhanced LCPUFAs biosynthesis



# CPT1A and Fatty Acid Oxidation

The CPT1A Arctic variant influences mitochondrial beta-oxidation of fatty acids. In high-fat, low-carbohydrate environments, enhanced fatty acid metabolism may confer energetic efficiency.

However, recent epidemiological evidence suggests potential trade-offs, including increased infant mortality under dietary transition conditions.

This illustrates a central evolutionary principle:

Adaptations are environment-specific and may become **maladaptive under ecological change.**

# KEY GENES FOR THE DETOXIFICATION

Pathway	Key Genes	Function	Arctic Relevance	Mediterranean Relevance
Phase I (Oxidation)	CYP1A1, CYP1A2, CYP1B1	Activation & biotransformation of POPs	Possible selection under high chronic exposure	Likely polymorphism-driven susceptibility differences
Aryl Hydrocarbon Receptor (AHR)	AHR	Regulates response to dioxins/PCBs	Important under extreme marine mammal diet	Important in polluted coastal urban zones
Phase II (Conjugation)	GSTM1, GSTT1, GSTP1	Glutathione conjugation & detox	May buffer chronic exposure	Null variants common → variable risk
Sulfation / Glucuronidation	UGT family	Enhance excretion	Relevant for lipid-soluble toxicants	Relevant but less intensively studied
Antioxidant System	SOD2, GPX1	Oxidative stress mitigation	Critical under high lipid peroxidation	Important in polluted harbors
Nuclear Receptors	PPAR $\alpha$ , PPAR $\gamma$	Lipid metabolism regulation	Interacts with high omega-3 intake	Interacts with Mediterranean lipid profile

Review

## Genetic Biomarkers of Metabolic Detoxification for Personalized Lifestyle Medicine

Lucia Aronica <sup>1,2,\*</sup>, Jose M. Ordovas <sup>3,4,5</sup>, Andrey Volkov <sup>6</sup>, Joseph J. Lamb <sup>7</sup> , Peter Michael Stone <sup>7,8,9,10</sup>, Deanna Minich <sup>8,11</sup> , Michelle Leary <sup>12</sup>, Monique Class <sup>8,13</sup>, Dina Metti <sup>7</sup> , Ilona A. Larson <sup>1</sup>, Nikhat Contractor <sup>14</sup>, Brent Eck <sup>1</sup> and Jeffrey S. Bland <sup>15</sup>

*Nutrients* **2022**, *14*, 768. <https://doi.org/10.3390/nu14040768>

Common genetic variants within genes involved in Phase I/Phase II detox reactions associated with **variability of response to foods or nutrients that modulate detox metabolism.**

Phase I Detox Enzymes		
<i>CYP1A2</i>		
Effect allele	Allele frequency	Effects on enzymatic function
rs762551-C Strength of evidence: Convincing (A).		C-allele carriers produce an enzyme variant with 62–70% lower activity and are less inducible by xenobiotics. Low CYP1A activity can result in decreased clearance of toxins, a lower 2/16-alpha hydroxyestrone ratio, and a higher risk of certain cancers. Consequently, lower production of reactive detoxification intermediates may reduce oxidative stress.
<i>CYP1B1</i>		
Effect allele	Allele frequency	Effects on enzymatic function
rs1056836-C Strength of evidence: Possible (C).		Individuals with the CC genotype tend to have higher enzymatic activity than G-allele carriers, which may result in greater activation of toxicants, greater production of 4-hydroxy estrogens, and greater oxidative damage. The effects of this SNP are affected by age, ethnicity, and menopausal status.

## Phase II detox enzymes

### *GSTM1*

Effect allele	Allele frequency	Effects on enzymatic function
<p><i>GSTM1</i> deletion  <i>GSTT1</i> deletion                      Strength of evidence:                      Probable (B).</p>	<p>→ - : 33-73%*                      → + : 27-67%                      → - : 33-73%*                      → + : 27-67%                      * -: deletion; +: present</p>	<p>Individuals carrying <i>GSTM1</i> or <i>GSTT1</i> double deletions (-/- genotype) may have a decreased ability to detoxify environmental toxicants, carcinogens, and products of oxidative stress. Gene deletions are more frequent among Caucasian and Asian populations and less frequent in African populations. Different segmental deletions have different frequencies in the population and between different ethnicities.</p>

### *COMT*

Effect allele	Allele frequency	Effects on enzymatic function
<p>rs4680-A                      Strength of evidence:                      Probable (B).</p>	<p>→ G: 63%                      → A: 37%</p>	<p>The A allele (Met) produces an enzyme with 40 % lower activity than that encoded by the G allele (Val). A-allele carriers may have a decreased ability to degrade neurotransmitters, estrogen, and various xenobiotics. This may result in increased sensitivity to environmental toxicants, a higher risk of developing neuropsychiatric disorders, and impaired estrogen metabolism.</p>

### *UGT1A1*

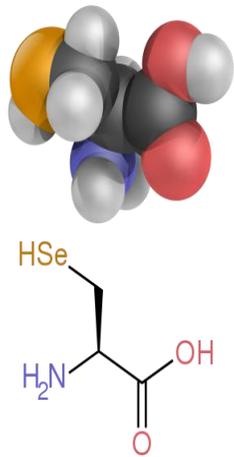
Effect allele	Allele frequency	Effects on enzymatic function
<p>rs3064744-TA                      Strength of evidence:                      Possible (C).</p>	<p>→ TA : 39%                      → - : 61%</p>	<p>Individuals carrying two insertion alleles (TA/TA genotype) may have a lower enzymatic activity than those carrying at most one copy of the deletion allele (-). This may result in increased toxicity in response to certain drugs (acetaminophen) and to a benign cardio-protective condition known as Gilbert syndrome, characterized by increased serum levels of total and unconjugated bilirubin.</p>

# Glutathione enzyme and selenoprotein polymorphisms

## TWO DIFFERENT ALLELE EFFECT

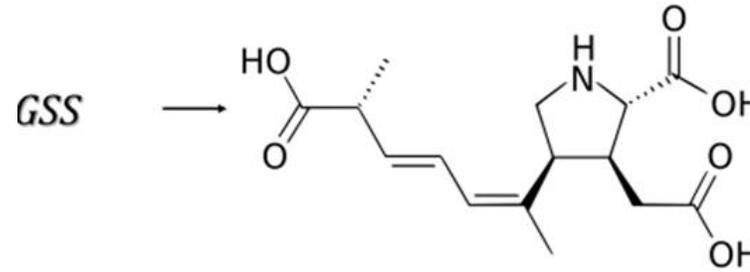
it encodes for an enzyme, glutathione synthetase, that is involved in the synthesis of glutathione, to which mercury is conjugated before being eliminated.

*SEPP1*



**Selenoprotein P  
plasma 1**

*SEPP1* 3'UTR T allele is linked to greater *SEPP1* expression and mercury binding capacity.



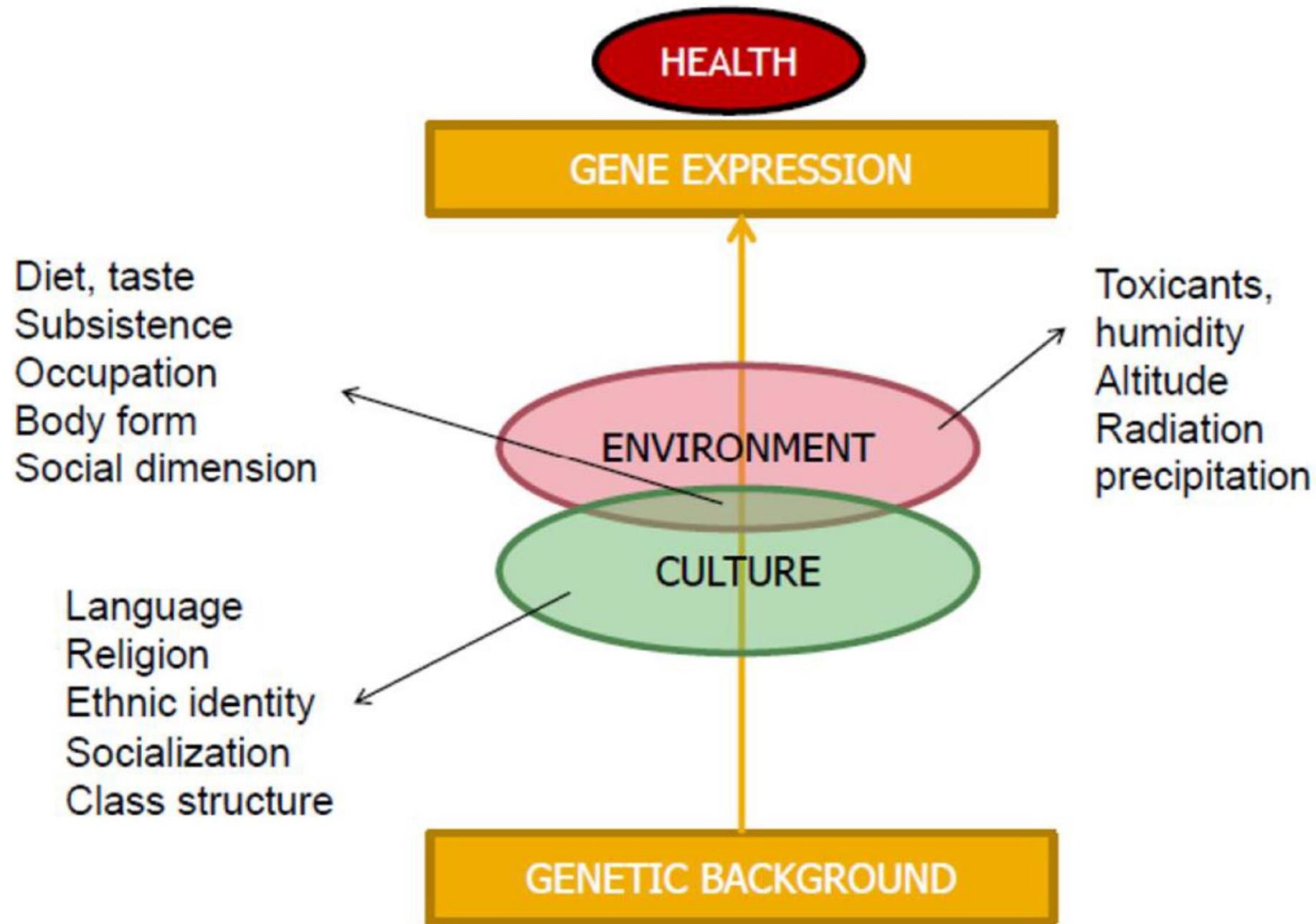
**Glutathione synthetase is  
a potent antioxidant**

Minor allele (G)

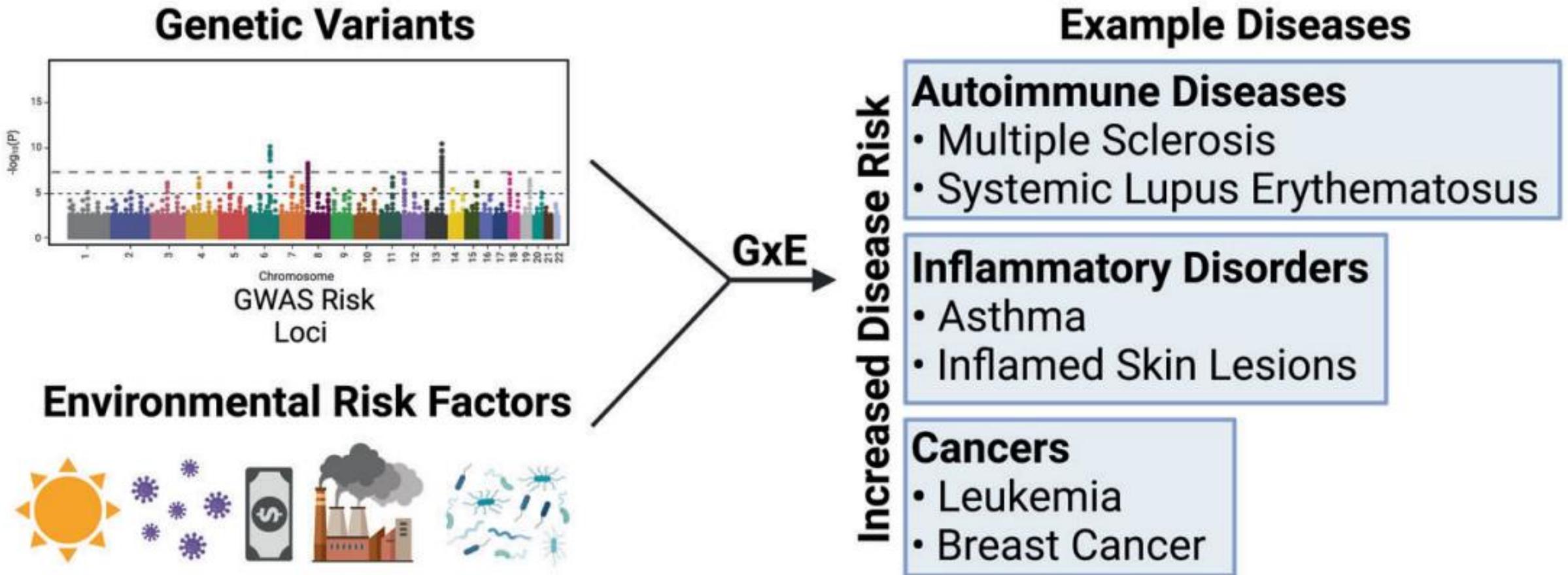
Decreased expression of *GSS*

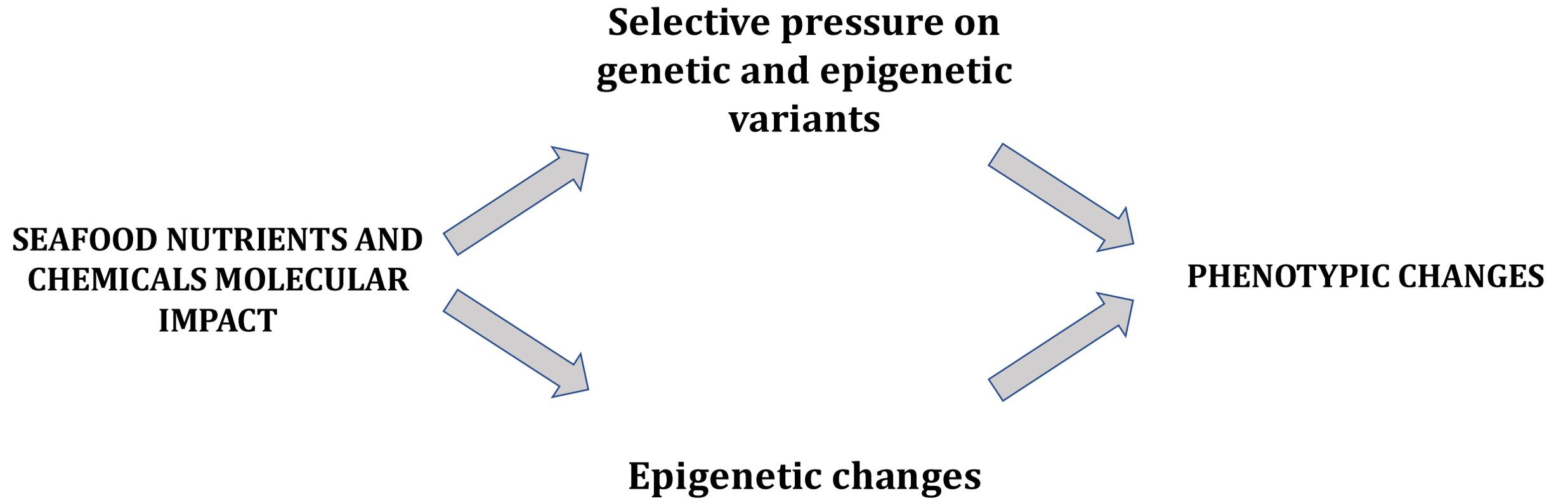
Reduced MeHg  
elimination

*SEPP1* encodes a selenoprotein, which fights the oxidative stress created by mercury by binding the toxicant directly via a selenocysteine residue



# Gene × environment (G × E) interactions involve synergy between environmental risk factors and genetic variants.

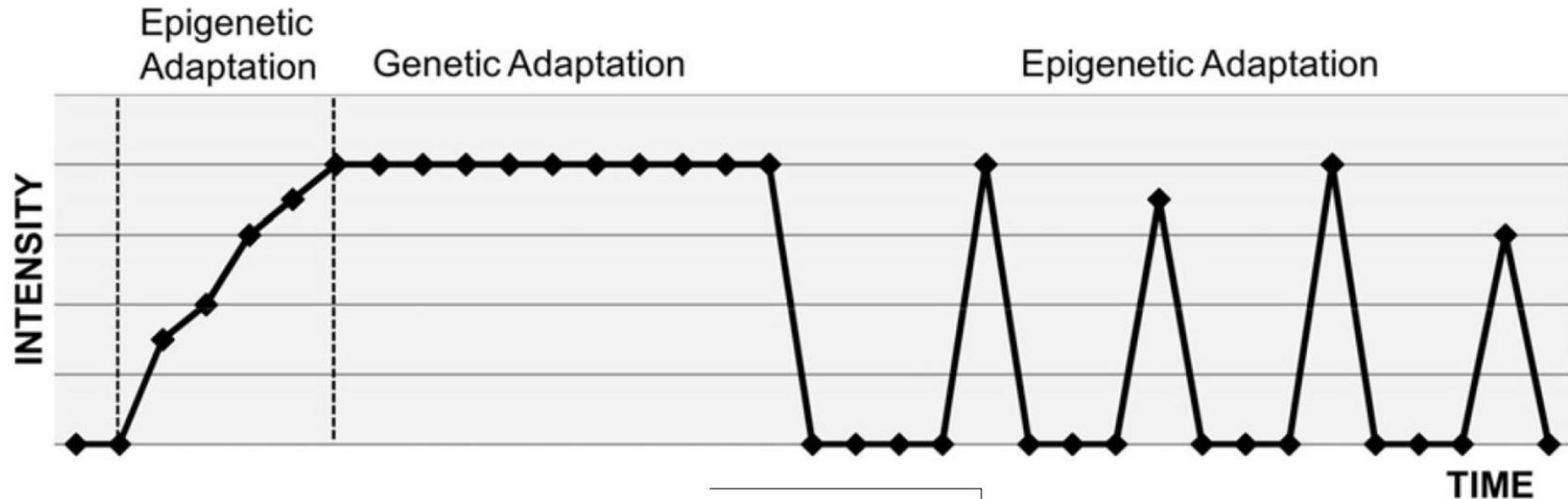




# EPIGENETIC SIDE OF HUMAN ADAPTATION

“When natural selection acts on pure epigenetic variation in addition to genetic variation, populations adapt faster, and adaptive phenotypes can arise before any genetic changes.” (Klironomos et al., 2013- Bioessay)

**STIMULUS (such as diet, pathogens, climate)**



Review Paper

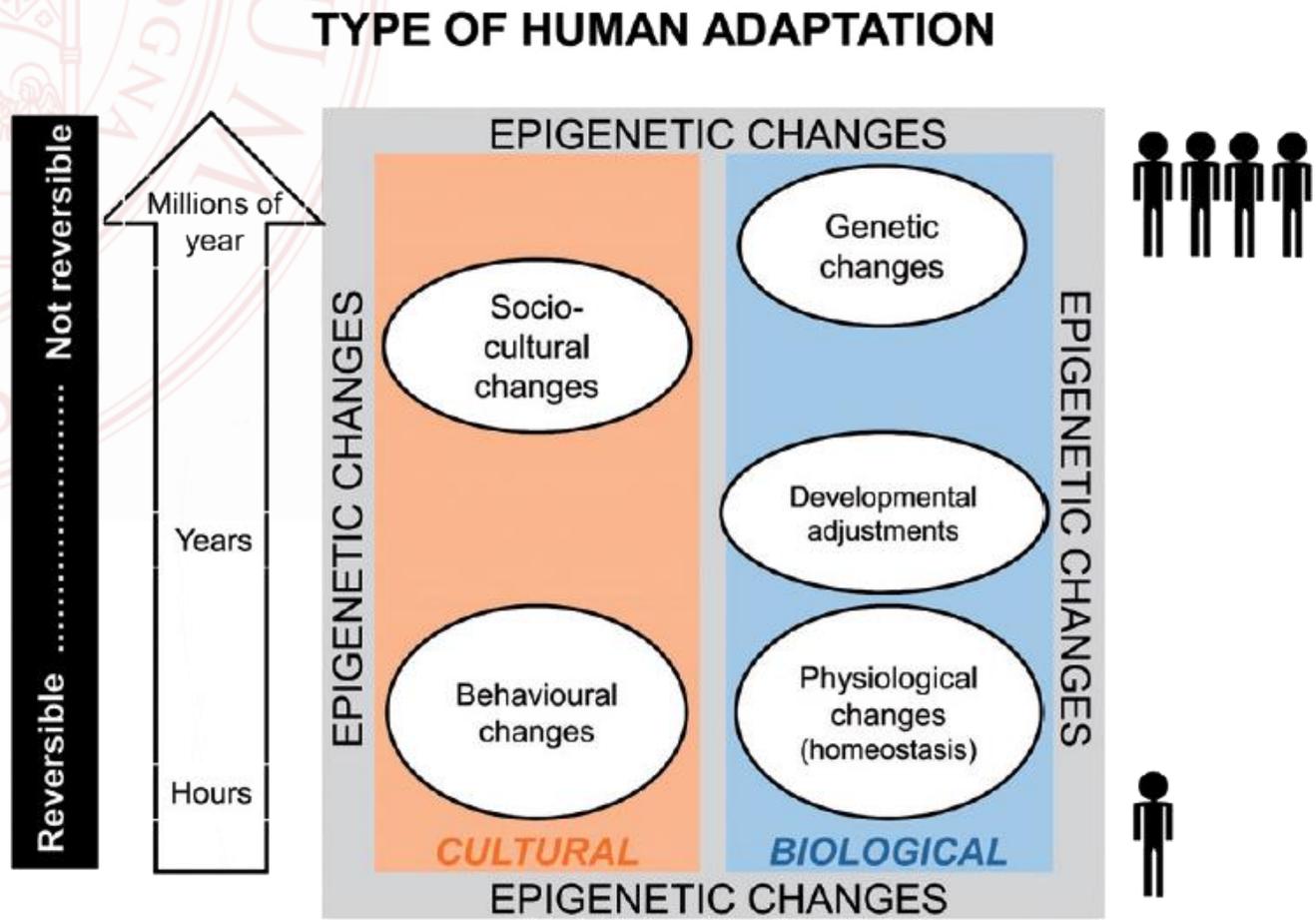
## The epigenetic side of human adaptation: hypotheses, evidences and theories

Cristina Giuliani, Maria Giulia Bacalini, Marco Sazzini, Chiara Pirazzini, Claudio Franceschi, Paolo Garagnani & Donata Luiselli ...show less

Pages 1-9 | Received 10 Jul 2014, Accepted 02 Sep 2014, Published online: 21 Nov 2014

# Giuliani et al., 2014. The epigenetic side of human adaptation: hypotheses, evidences and theories

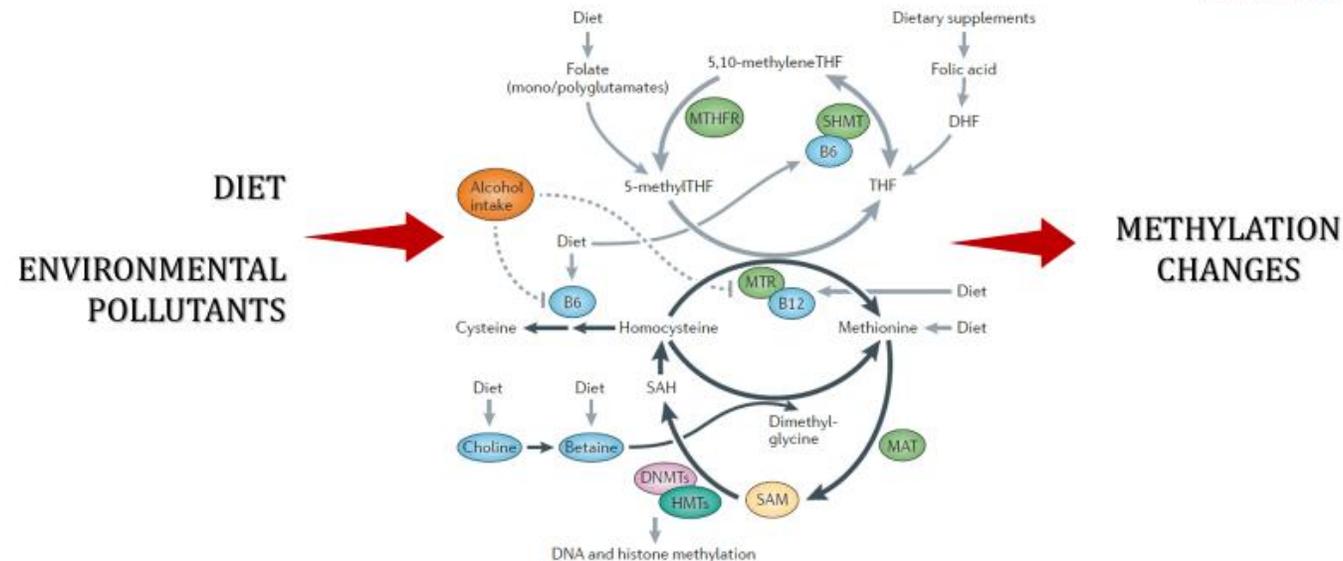
Epigenetics mechanisms could represent “medium-term” strategies to cope with a demanding environmental condition



Epigenetic changes  Gene expression changes

**A common type of epigenetic modification is called DNA methylation.** DNA methylation involves the attachment of small chemical groups called methyl groups (each consisting of one carbon atom and three hydrogen atoms) to DNA building blocks

### Synthesis of methylation enzymes

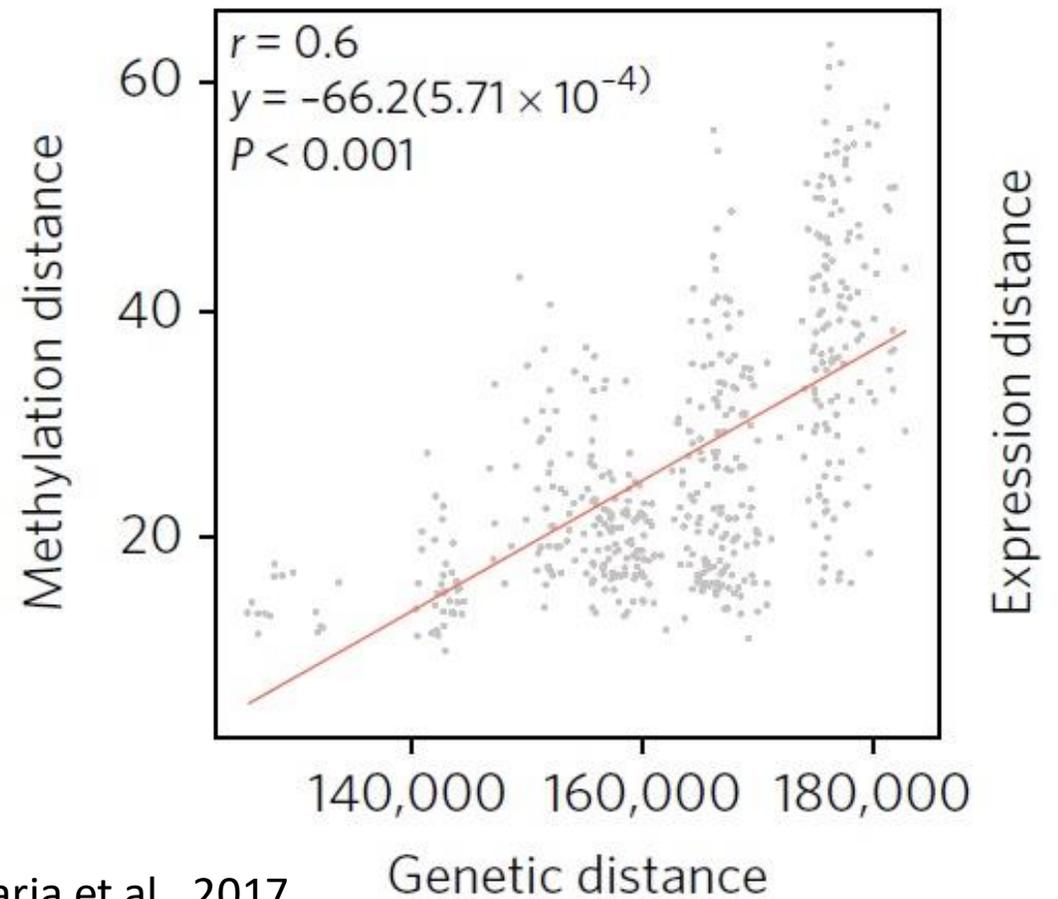


# Methylation variability across human populations

Environment

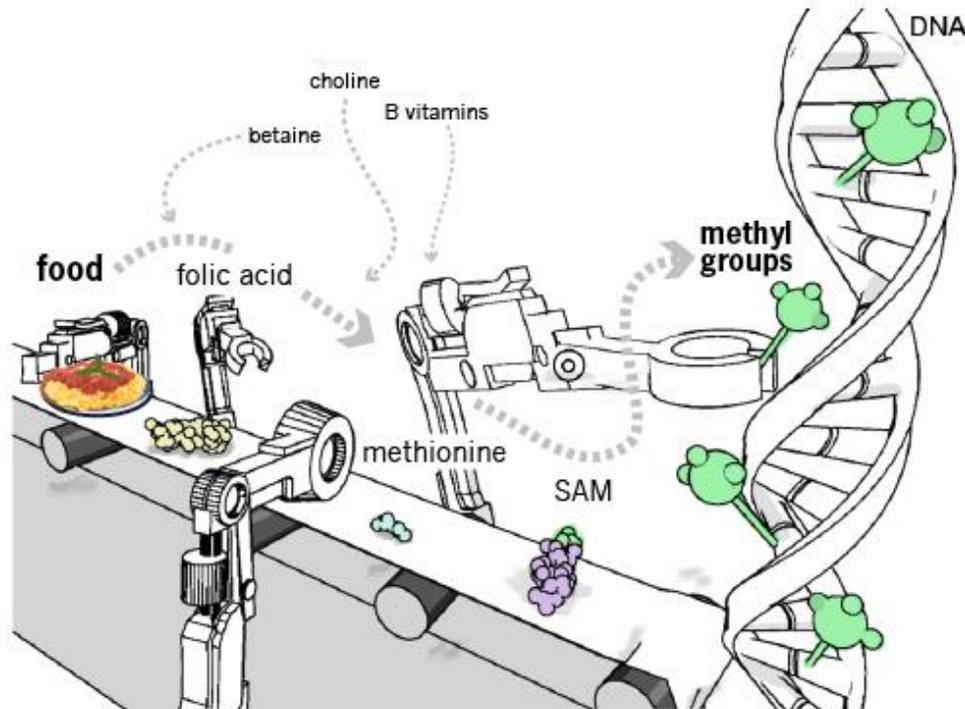
- Diet
- UVA exposure
- Pathogens load
- ...

Genetic background



Could methylation changes contribute to human adaptation?

# EPIGENETICS and DIET



*Nutrients from our food are funneled into a biochemical pathway that extracts methyl groups and then attaches them to our DNA.*

Unlike behavior or stress, diet is one of the most easily studied, and therefore best understood, environmental factors in epigenetic change. The nutrients we extract from food enter metabolic pathways where they are manipulated, modified, and molded into molecules that the body can use. One of these pathways is responsible for the creation of methyl groups, important epigenetic tags capable of modulating gene expression

# EPIGENETICS and DIET



The foods we eat can be considered real epigenetic modulators capable of influencing the development and maintenance of the body.

Nutrient molecules, in addition to being metabolic modulators, have a direct correlation with DNA.

Different people respond very differently to the same foods by virtue of their genetic background, but it has also been seen that the foods themselves can influence DNA expression.

# Conclusions

Traditional fishing communities illuminate how human populations adapt to specific ecological niches through **genetic, nutritional, and cultural pathways**. These adaptations are context-dependent and increasingly destabilized by **globalization and climate change**.

A **multi-layered evolutionary system**, where biological and cultural processes interact across timescales.

Coastal adaptation is not reducible to simple genetic change. Instead, it emerges from the interaction of **three evolutionary dynamics**:

- 1. Genetic adaptation** (Darwinian selection on heritable variation)
- 2. Phenotypic plasticity** (developmental and physiological flexibility)
- 3. Cultural evolution** (technological and social buffering mechanisms)

# Integrated Systems Model

We can conceptualize coastal adaptation as a three-layer system:

## Environmental Pressures

(diet, salt, pathogens, hypoxia, temperature)



## Immediate Response: Phenotypic Plasticity



## Medium-Term Buffering: Cultural Innovation



## Long-Term Response: Genetic Adaptation



Modified Population Phenotype

These layers are not sequential, but interactive.

For example:

- Cultural buffering reduces mortality.
- Reduced mortality alters selection gradients.
- Plastic responses expose new phenotypes to selection.
- Genetic adaptation modifies plastic capacity.

# Temporal Scale Perspective

Process	Timescale	Mechanism
Plasticity	Days–Years	Physiological regulation
Cultural evolution	Years–Centuries	Social learning
Genetic adaptation	Generations–Millennia	Allele frequency change

Coastal adaptation must therefore be analyzed as a **nested evolutionary system across timescales**.

These processes operate simultaneously, but on different temporal and mechanistic levels. These adaptations are not universally protective, but contingent upon ecological continuity.

Rapid modernization reveals the fragility of tightly coupled gene–environment systems.

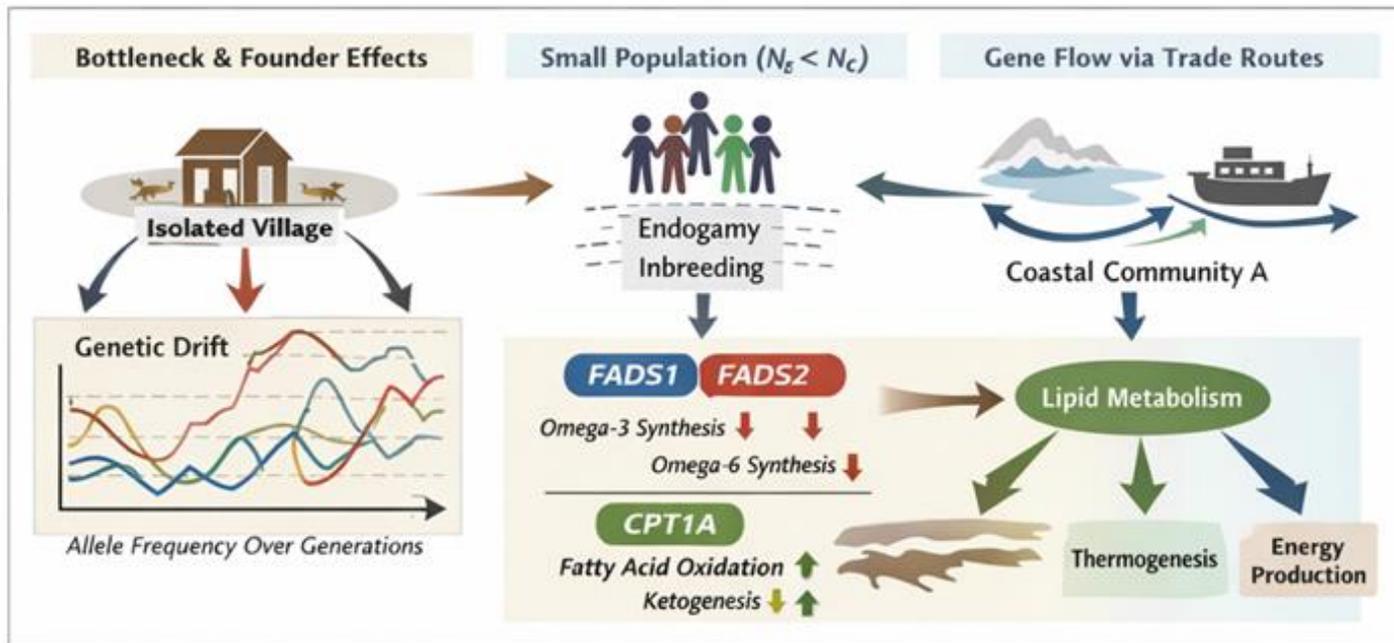
Integrating evolutionary biology with public health and environmental science is essential for developing ethically grounded, sustainable interventions.

## Key Insight

Human coastal adaptation is not simply biological adaptation to the sea. It is a dynamic interaction between:

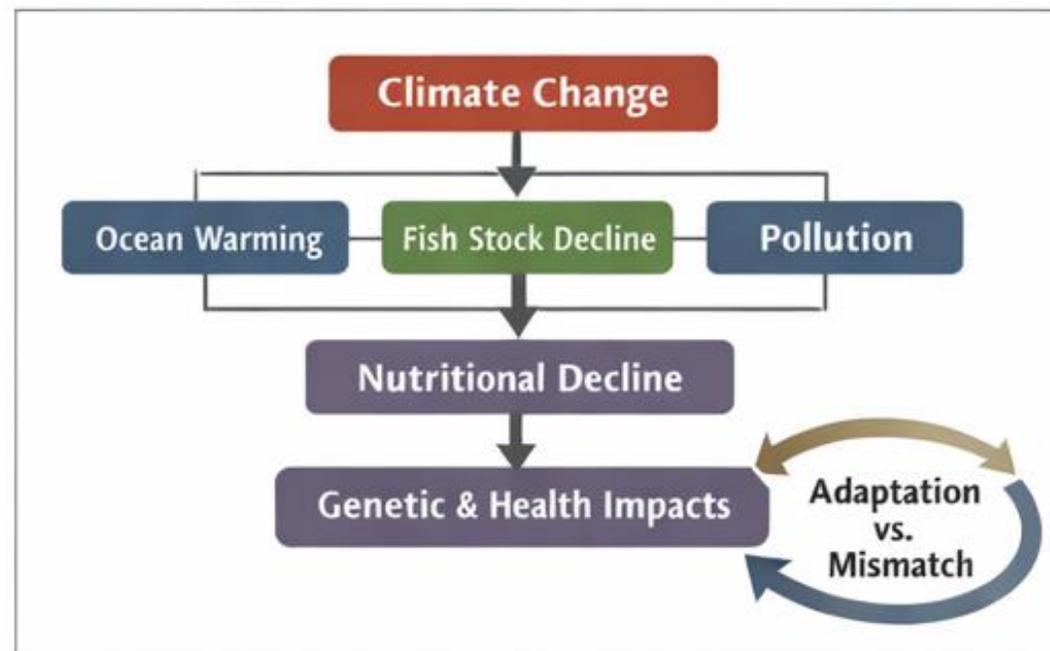
- Ecological constraints
- Physiological flexibility
- Cultural innovation
- Genetic evolution

It represents one of the clearest examples of how humans evolve not only biologically, but also culturally and developmentally in complex ecosystems.



## Graphical conclusions

### Climate Change and Marine Food Systems





# Thank you for your attention



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## **I. Genetic Adaptation**

*(Long-term evolutionary response)*

Genetic adaptation occurs when:

- There is heritable variation in a trait.
- Environmental conditions create differential survival or fertility.
- Advantageous alleles increase in frequency over generations.

In coastal environments, selective pressures may include:

- Marine-based diets rich in omega-3 fatty acids and iodine
- Chronic salt exposure
- Hypoxic diving stress
- Pathogen-dense wetlands
- Thermal stress (humid tropics or Arctic cold coasts)

## Example of Selective Targets

- Lipid metabolism genes (marine fat consumption)
- Thyroid regulation (iodine-rich diets)
- Immune system genes (wetland pathogens)
- Hypoxia-response pathways (free diving populations)

Importantly, many coastal adaptations likely involve **polygenic selection**, meaning small shifts in allele frequency across many loci rather than dramatic single-gene sweeps.

## II. Phenotypic Plasticity

*(Within-lifetime flexibility)*

Phenotypic plasticity allows individuals to adjust to environmental conditions without genetic change.

Examples in coastal settings:

- Increased sweating efficiency in humid tropics
- Short-term blood pressure modulation under salt load
- Enlarged spleen response after repeated diving
- Microbiome shifts due to marine diet

Plasticity is crucial because:

- Coastal environments are highly variable (seasonal fish supply, storms, salinity shifts).
- Rapid environmental change may outpace genetic evolution.

Plastic responses can later become genetically stabilized through **genetic accommodation** if selection consistently favors certain reaction norms.

### III. Cultural Evolution

*(Rapid adaptive buffering system)*

Humans uniquely modify their selective environment through culture.

In coastal contexts:

- Fishing technologies
- Boats and navigation systems
- Salt extraction techniques
- Food preservation (drying, fermentation)
- Water filtration systems

Cultural practices reduce direct environmental pressure.

This creates a feedback loop:

Environment

- Cultural innovation
- Reduced biological stress
- Altered selection intensity

This dynamic is known as **gene–culture coevolution**.

## **Multidimensional Selective Pressures in Coastal Environments**

Coastal ecosystems are ecotones — transitional zones between land and sea — which increases environmental complexity.

### **1. Marine-Based Diet**

Characteristics:

- High omega-3 polyunsaturated fatty acids
- High protein
- Elevated iodine
- Variable carbohydrate availability

Selective implications:

- Lipid metabolism regulation
- Thyroid hormone modulation
- Insulin pathway adjustments

### **2. Salinity and Osmotic Stress**

Exposure sources:

- Brackish water
- Salt-rich seafood
- High evaporative loss in humid heat

Physiological systems under pressure:

- Kidney sodium reabsorption
- Renin–angiotensin–aldosterone system
- Blood pressure homeostasis

Selection may favor individuals who:

- Maintain cardiovascular stability under salt load
- Avoid salt-sensitive hypertension