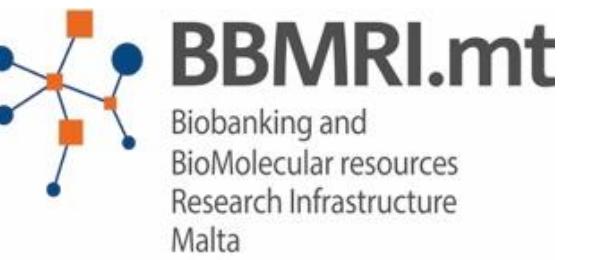


L-Università  
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# The University of Malta Biobank

## BBMRI.mt

Alex Felice  
Joanna Vella



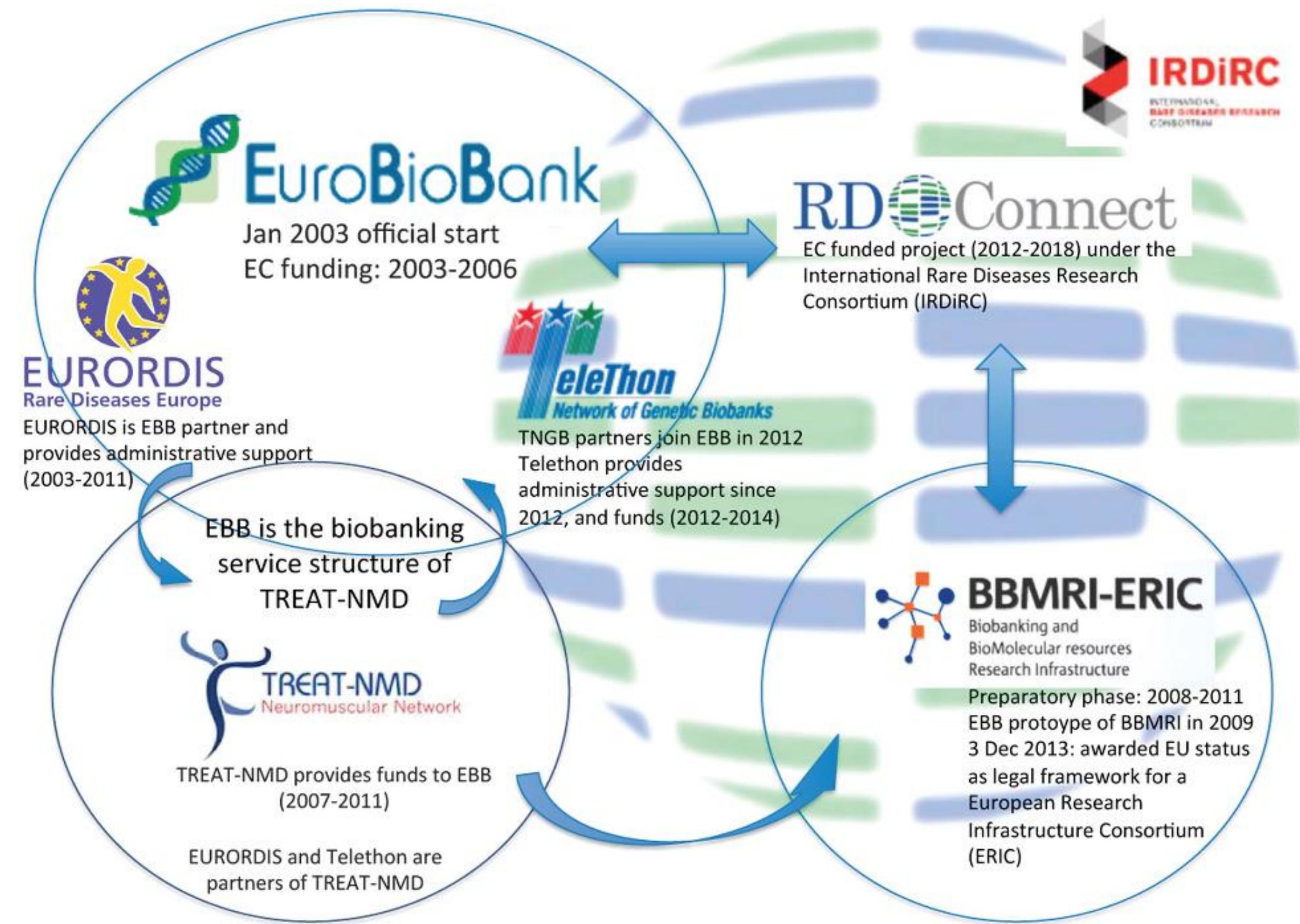
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# THE MALTA BIOBANK IS 32 YEARS YOUNG

- 1989: WHO & DH Hb Neonatal testing & Thalassaemia Clinic @ SLH/MDH; GlobinBank @ UM
- 1992: EU-FP AVICENNE - Research
- 1996: Malta Gov. / IPA Biotech – Geo-Parkinson
- 1998: Molecular Genetics Diagnostic Services, Rare Diseases
- 2000: Atheneum Biotechnology
- 2002: EuroBioBank
- 2008: Malta Gov. - T2DM Genomics
- 2009: ERDF & ESF - Enhanced Biotechnology
- 2013: BBMRI - ERIC & RD-CONNECT
- 2014: Centre for Molecular Medicine & Biobanking @ UM;  
MCST: The Maltese Genome Project
- 2015: ADOPT BBMRI-ERIC
- 2016: BBMRI.mt Euro-Mediterranean Engagement







npg

European Journal of Human Genetics (2015) 23, 1116–1123  
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## POLICY

# The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases

Marina Mora<sup>1\*</sup>, Corrado Angelini<sup>2,3</sup>, Fabrizia Bignami<sup>4</sup>, Anne-Mary Bodin<sup>5</sup>, Marco Crimi<sup>6</sup>, Jeanne Hélène Di Donato<sup>7</sup>, Alex Felice<sup>8</sup>, Cécile Jaeger<sup>9</sup>, Veronika Karcagi<sup>10</sup>, Yann LeCam<sup>5</sup>, Stephen Lynn<sup>11</sup>, Marija Meznaric<sup>12</sup>, Maurizio Moggio<sup>13</sup>, Lucia Monaco<sup>6</sup>, Luisa Politano<sup>14</sup>, Manuel Posada de la Paz<sup>15</sup>, Safaa Saker<sup>16</sup>, Peter Schneiderat<sup>17</sup>, Monica Ensini<sup>11</sup>, Barbara Garavaglia<sup>18</sup>, David Gurwitz<sup>19</sup>, Diana Johnson<sup>20</sup>, Francesco Muntoni<sup>20</sup>, Jack Puymirat<sup>21</sup>, Mojgan Reza<sup>1</sup>, Thomas Voit<sup>22</sup>, Chiara Baldo<sup>23</sup>, Franca Dagna Bricarelli<sup>24</sup>, Stefano Goldwurm<sup>25</sup>, Giuseppe Merla<sup>26</sup>, Elena Pegoraro<sup>3</sup>, Alessandra Remieri<sup>27</sup>, Kurt Zatloukal<sup>28</sup>, Mirella Filocamo<sup>29</sup> and Hanns Lochmüller<sup>11</sup>

The EuroBioBank (EBB) network ([www.eurobiobank.org](http://www.eurobiobank.org)) is the first operating network of biobanks in Europe to provide human DNA, cell and tissue samples as a service to the scientific community conducting research on rare diseases (RDs). The EBB was established in 2001 to facilitate access to RD biospecimens and associated data; it obtained funding from the European Commission in 2002 (5th framework programme) and started operation in 2003. The set-up phase, during the EC funding period 2003–2006, established the basis for running the network; the following consolidation phase has seen the growth of the network through the joining of new partners, better network cohesion, improved coordination of activities, and the development of a quality-control system. During this phase the network participated in the EC-funded TREAT-NMD programme and was involved in planning of the European Biobanking and Biomolecular Resources Research Infrastructure. Recently, EBB became a partner of RD-Connect, an FP7 EU programme aimed at linking RD biobanks, registries, and bioinformatics data. Within RD-Connect, EBB contributes expertise, promotes high professional standards, and best practices in RD biobanking, is implementing integration with RD patient registries and 'omics' data, thus challenging the fragmentation of international cooperation on the field.

*European Journal of Human Genetics (2015) 23, 1116–1123; doi:10.1038/ejhg.2014.272; published online 24 December 2014*

## INTRODUCTION

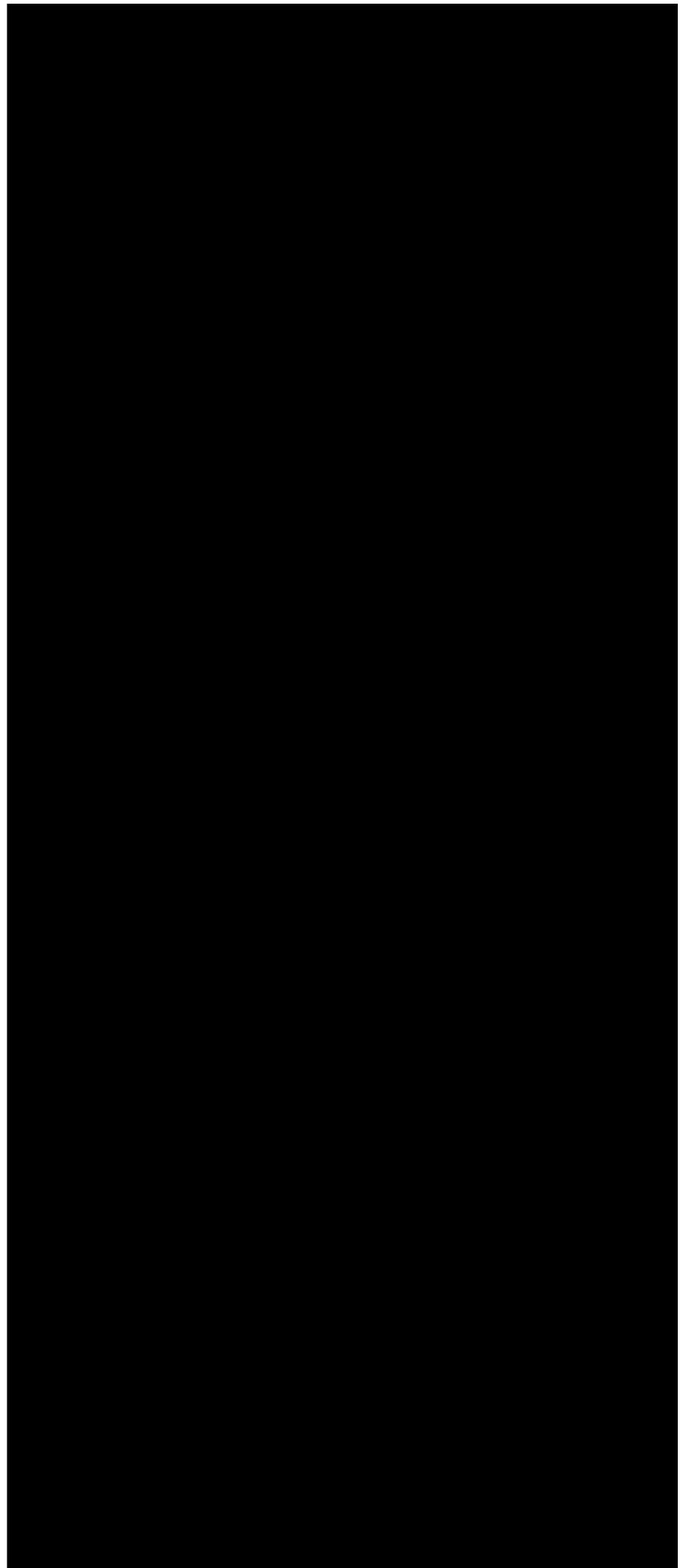
The Organisation for Economic Co-operation and Development (OECD) defines a biobank as 'A collection of biological material and the associated data and information stored in an organised system, for a population or a large subset of a population'.<sup>1</sup> The collection of biological material and data for research and diagnosis has a long history in educational and medical institutions. In the past,

biorepositories tended to be inconspicuous—the responsibility of individual research groups or institutions, and biospecimens were rarely shared with other laboratories. With recent technological advances, biorepositories are being opened up for new uses (when permitted by national regulations), and new biorepositories are being established as part of funded, but time-limited, research projects, whereas information technology now enables the systematic linkage

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or Professor H Lochmüller, Chair of Experimental Myology, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, Newcastle University, Newcastle-upon-Tyne NE1 3BZ, UK. Tel: +44 191 2418602; Fax: +44 191 2418770; E-mail: hanns.lochmuller@ncl.ac.uk

Received 11 August 2014; revised 15 October 2014; accepted 10 November 2014; published online 24 December 2014



# THE UNIVERSITY OF MALTA BIOBANK

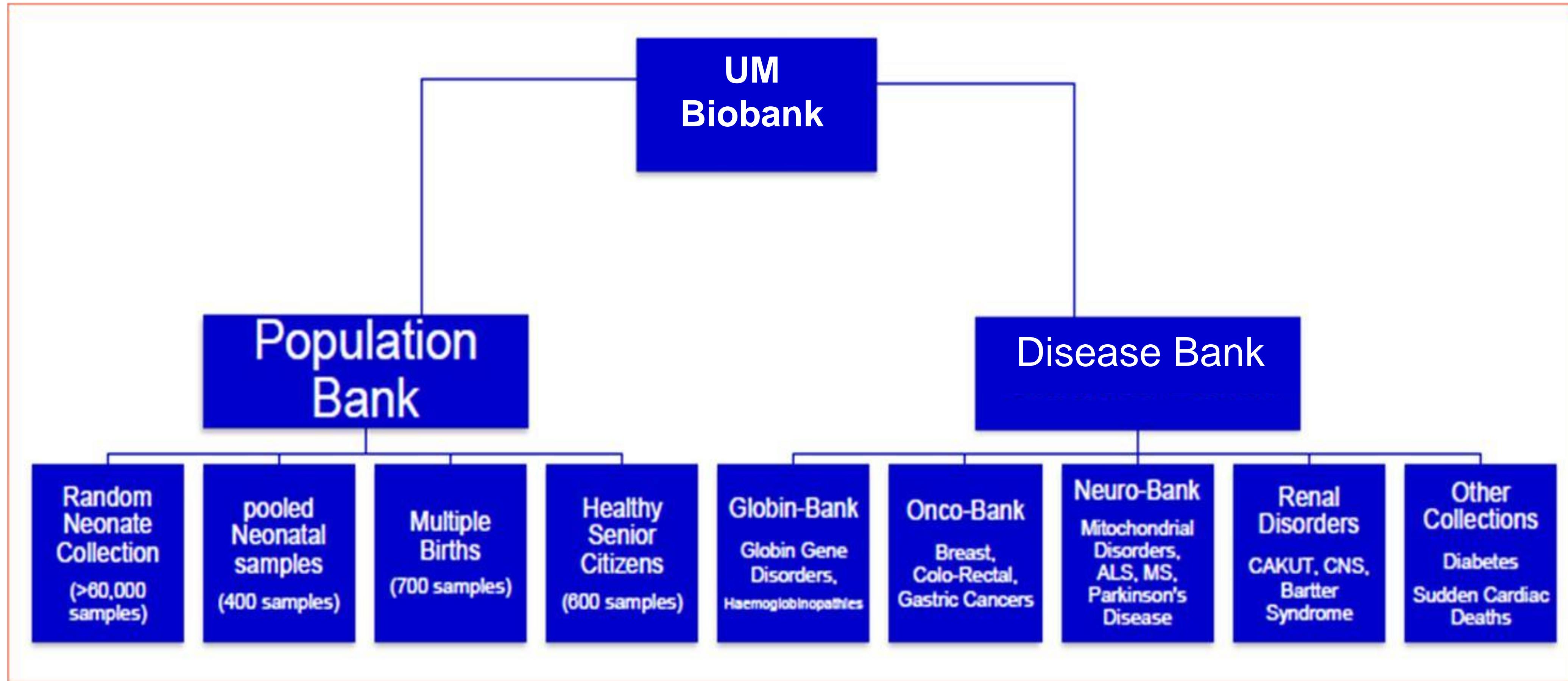
## = BBMRI.mt

SPECIFIC OBJECTIVE;

o TO CONSTRUCT HIGH QUALITY TOOLS FOR DISCOVERY RESEARCH WITH WELL ORGANISED INTER-OPERABLE COLLECTIONS OF HUMAN SAMPLES AND HEALTH DATA OVER TIME WITH RESEARCH PARTNERSHIPS AT NATIONAL, REGIONAL AND EUROPEAN LEVELS.

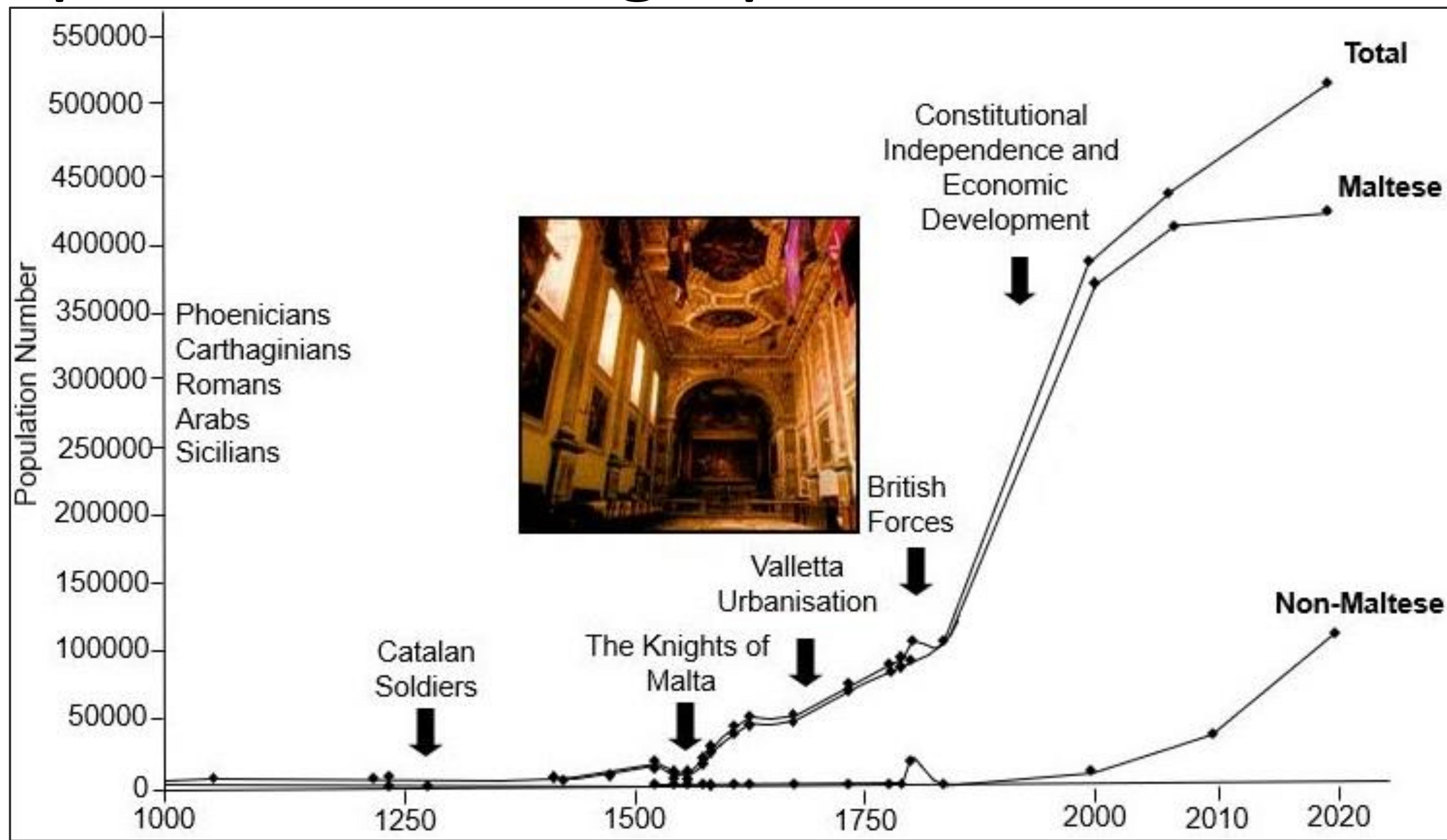
o LONG TERM GOALS (RARE DISEASE)  
BIOBANK EMBEDMENT OF NEWBORN TESTING.  
BIOBANK LED (GENE DISCOVERY) RESEARCH.  
EXPEDITED DIAGNOSIS & NEW TREATMENTS.

# Malta's first national archive of blood and DNA samples



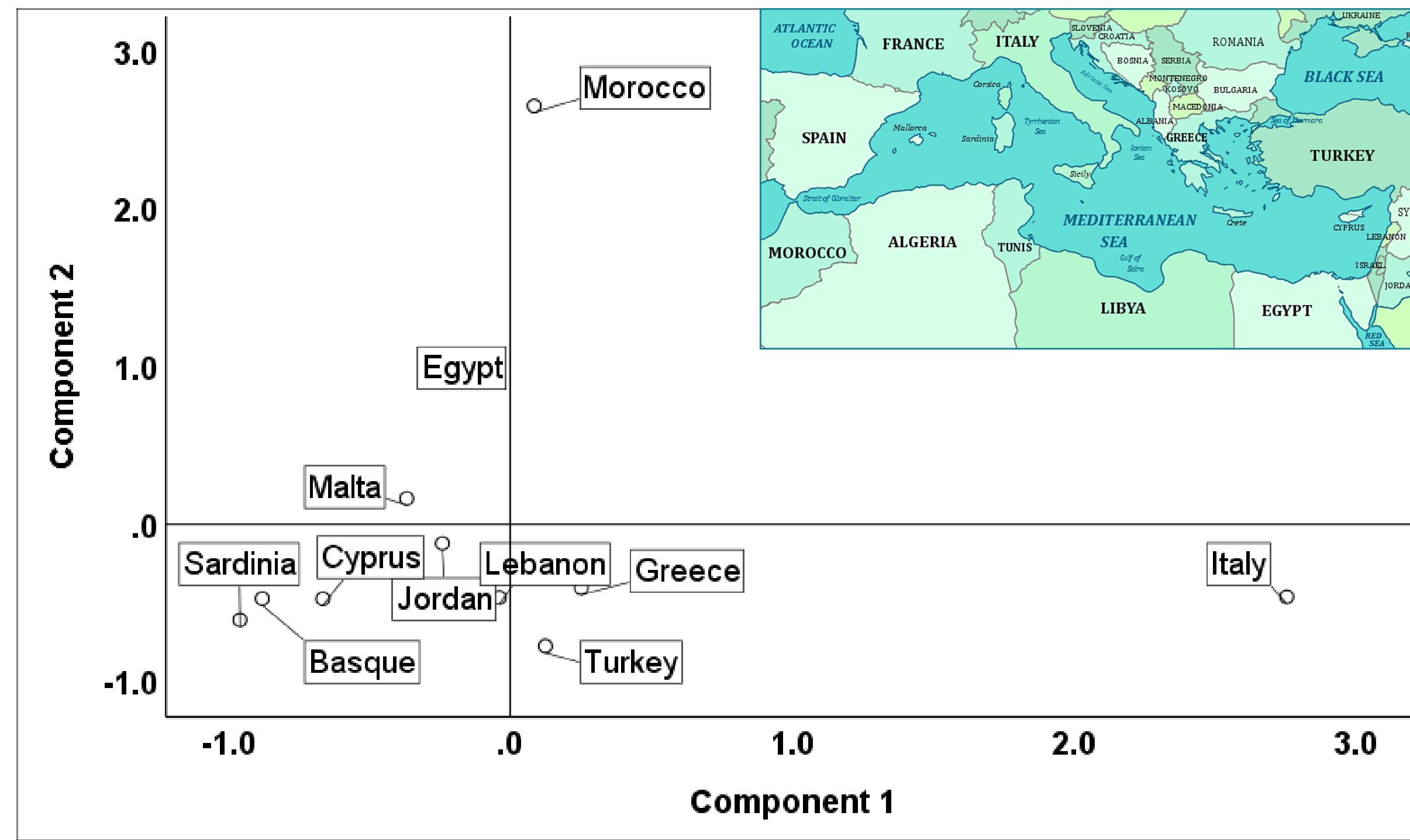
# Population Genetics

## Population Demographics of the Maltese



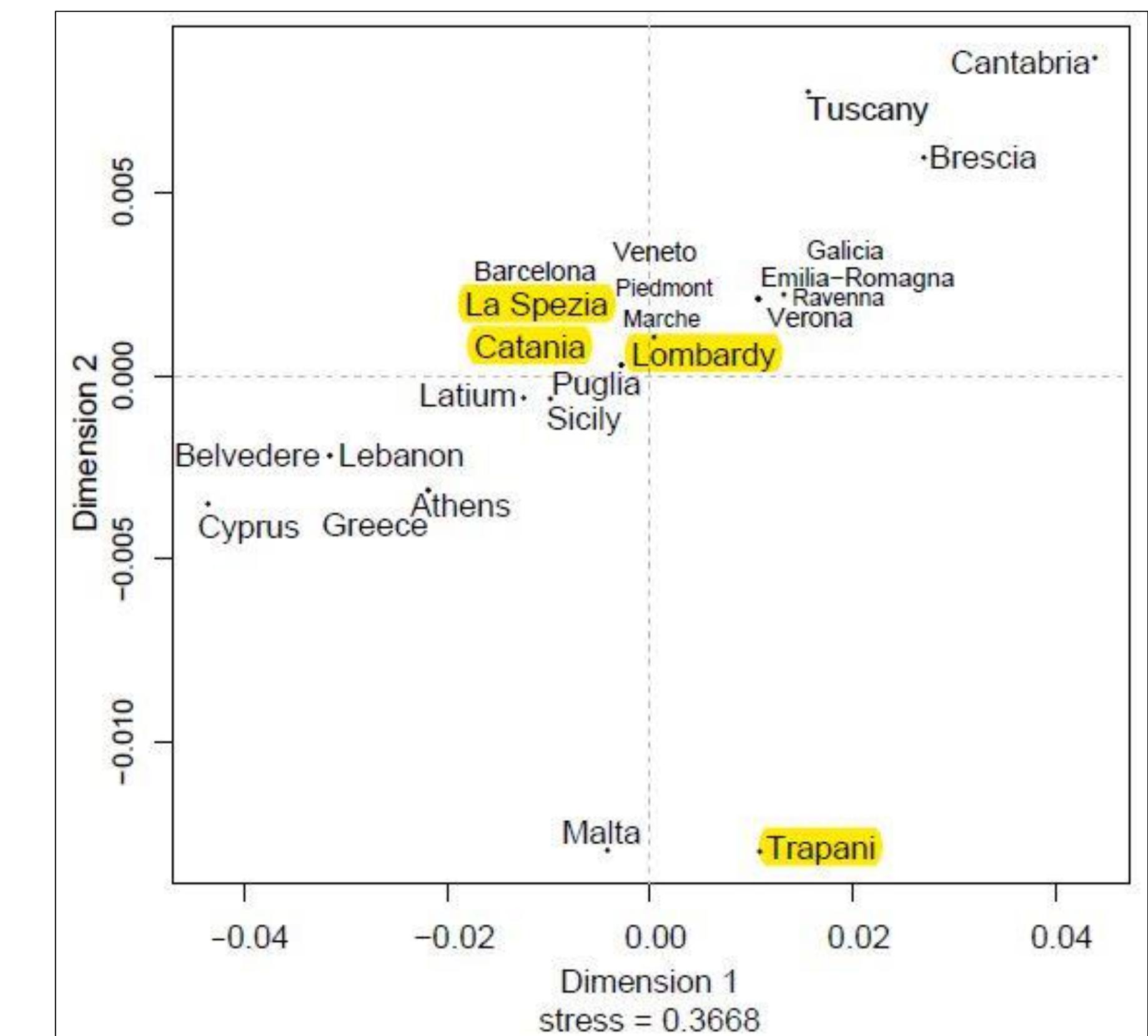
# Population Results: mtDNA and Y chromosome

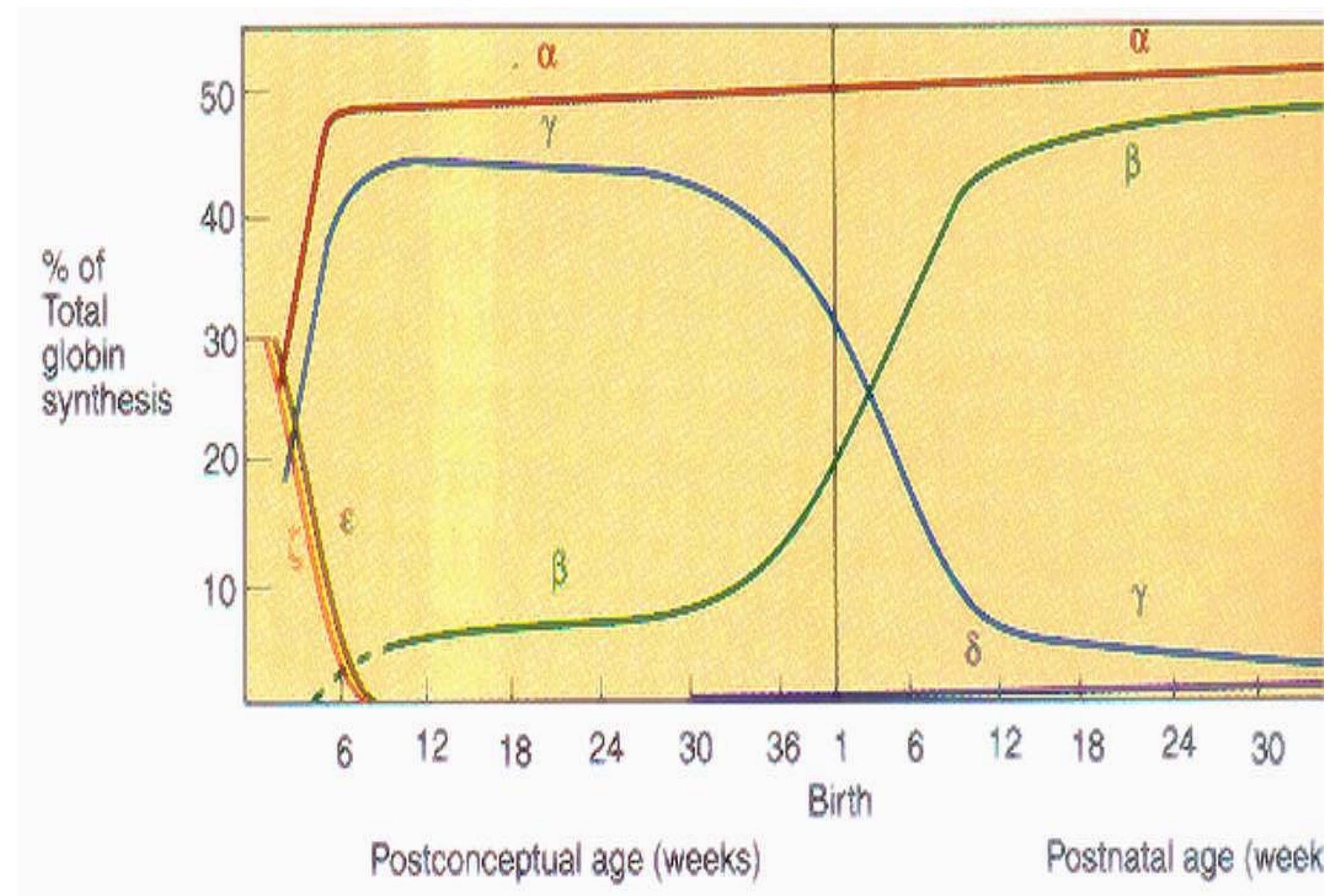
**Figure 1: PCA Plot loading scores  
of mtDNA haplogroup frequencies**

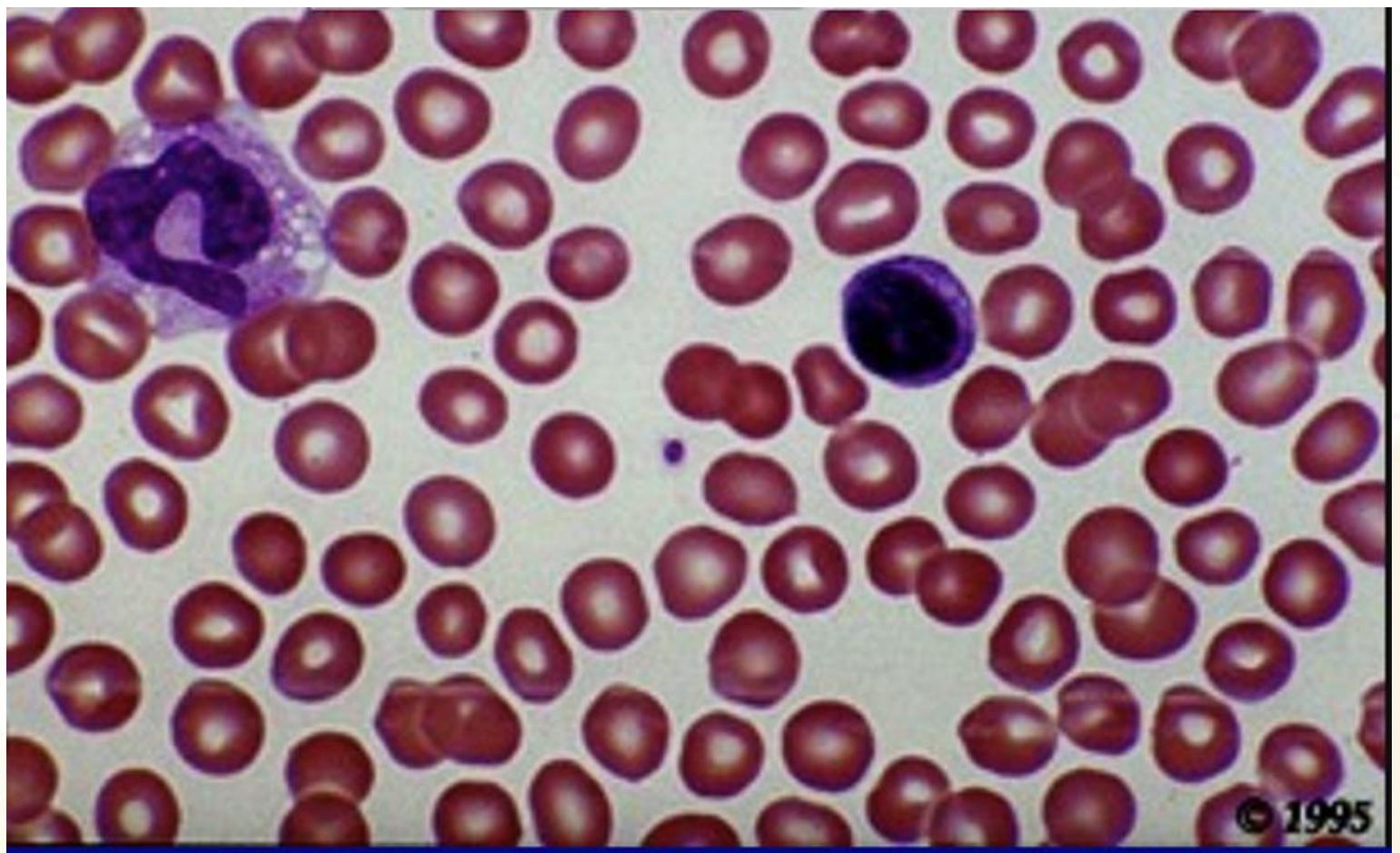


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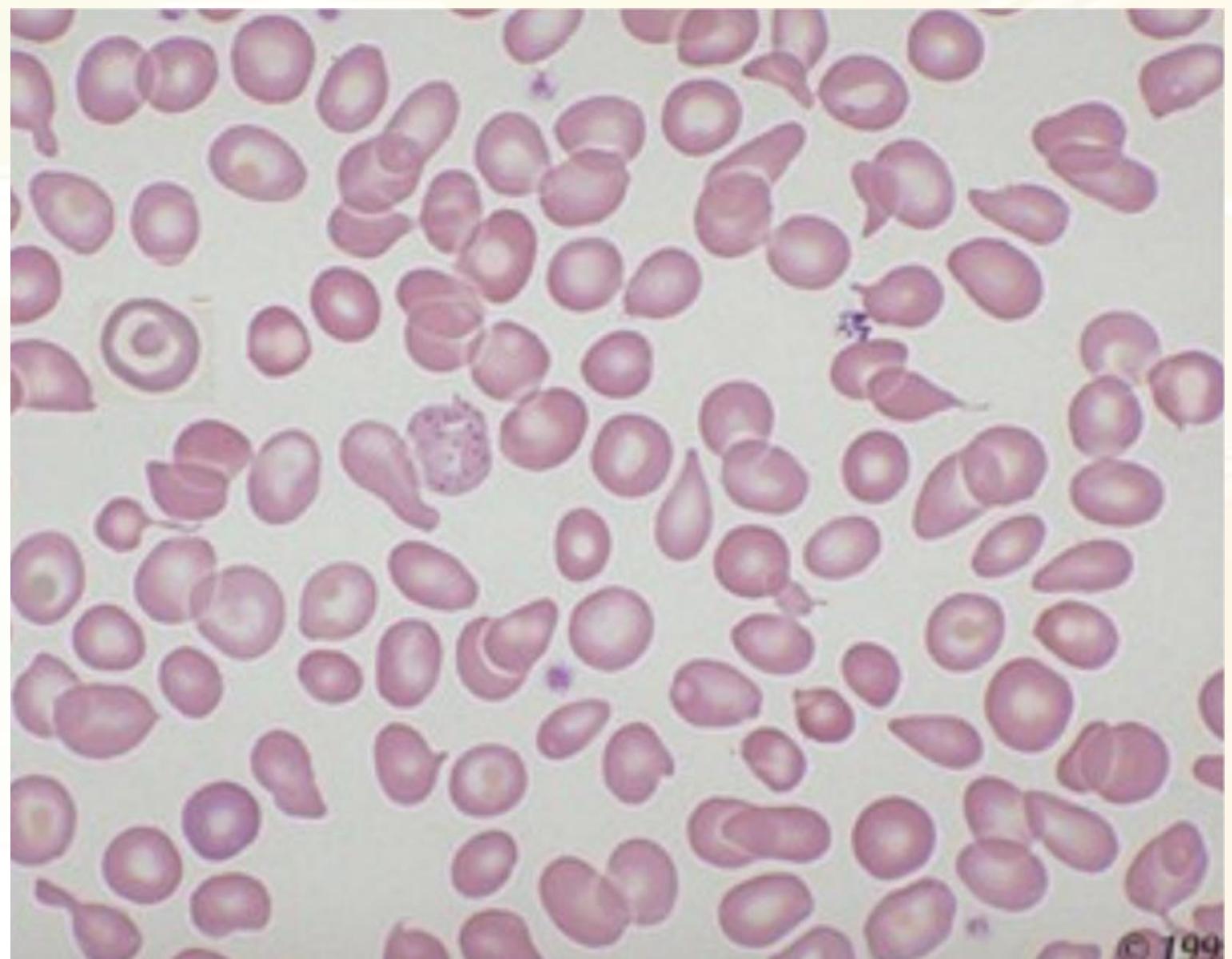
**Figure 2: MDS Plot AMOVA ( $R_{ST}$ )  
Maltese and other Mediterranean  
populations Y23 STR haplotypes**



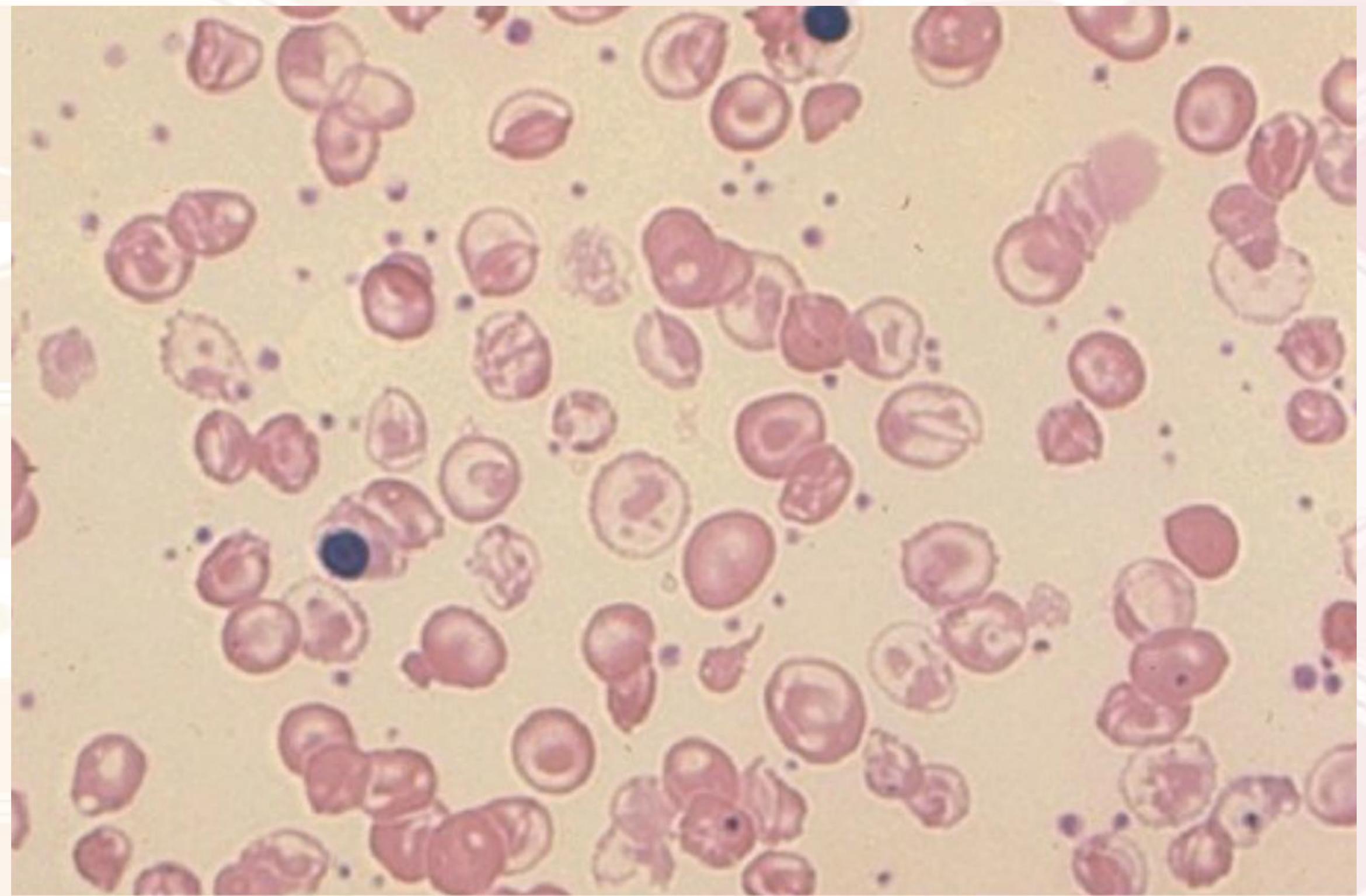




Normal (Hb AA)

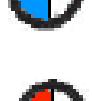
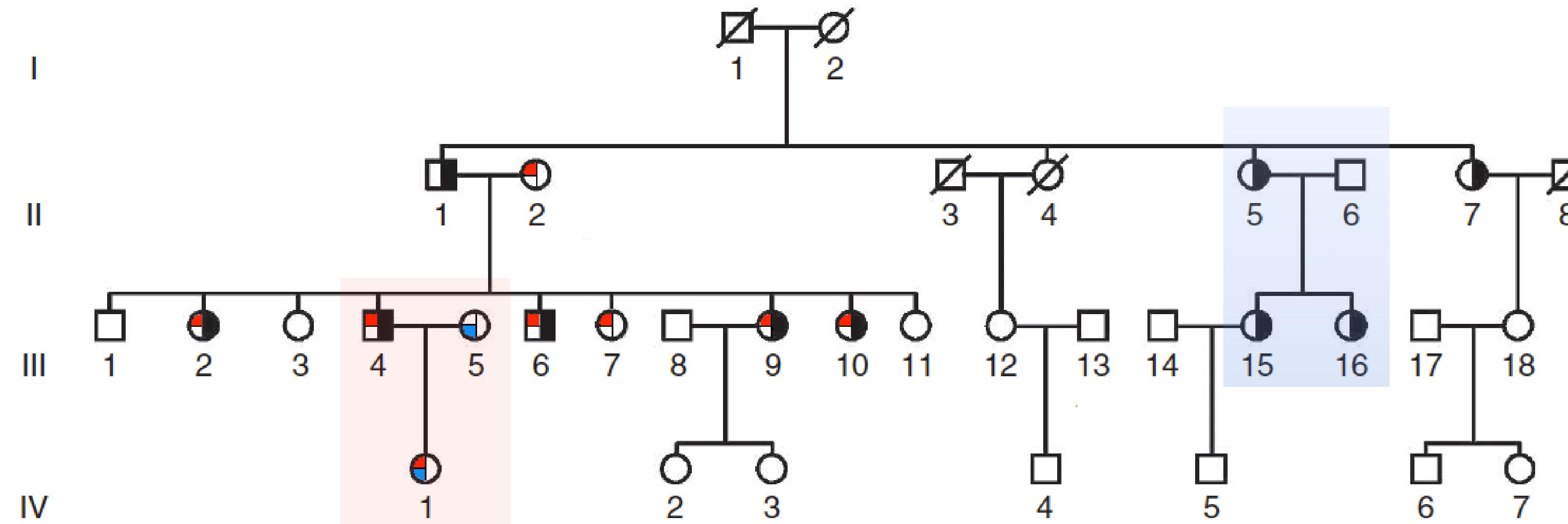


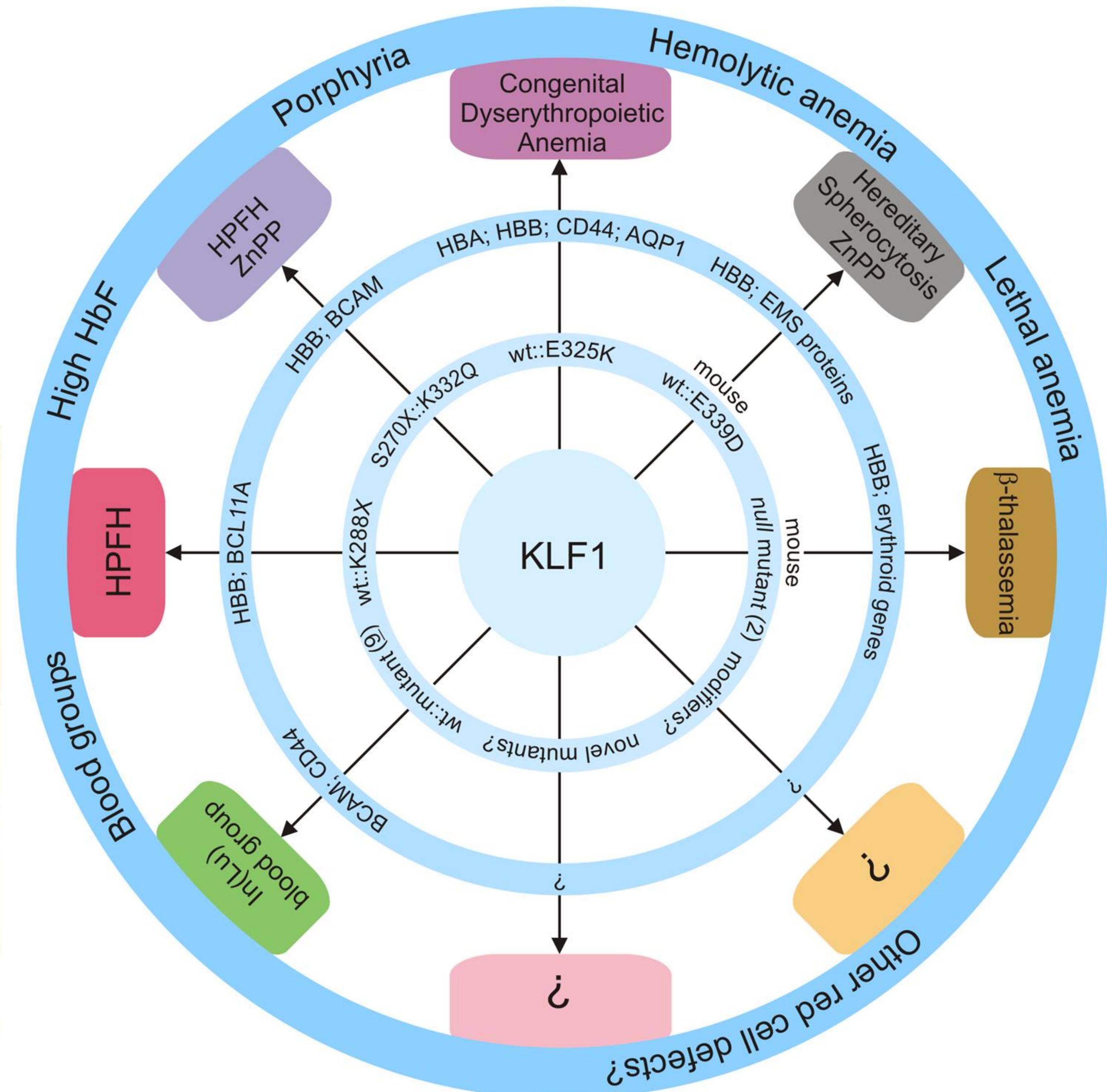
Thalassaemia Heterozygote (carrier)  
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Thalassaemia Homozygote (major)

# Family with KLF1 Haplo-insufficiency and HPFH (Malta Type)





## **CHALLENGES > 2022**

**FOCUS / VIRTUAL BIOBANKS.**

**EDUCATION & TRAINING.**

**AWARENESS.**

**THE TRUST ENVIRONMENT.**

**PUBLIC & PRIVATE ENGAGEMENT.**

**BIG DATA > SAMPLES (1+MG)**

**ACADEMY OF MEDICAL RIs**

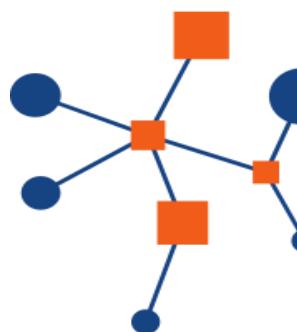




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RD Connect



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Biobanking and  
BioMolecular resources  
Research Infrastructure  
Malta