21ST

PGDIS CONFERENCE

PREIMPLANTATION GENETIC DIAGNOSIS INTERNATIONAL SOCIETY

PGDIS

6-8 May 2024

Kuala Lumpur

Malaysia

PGT and BEYOND...

21ST PGDIS CONFERENCE

PGT-M reproductive testing as a genetic disease prevention program

Dr Karolina Kobus

Rare Genetic Diseases – Global Perspective

Epidemiology

- Around 5 % all live births have genetic disease
- 7 million people are born with genetic disease per year
- 7000 genetic disorders

Challenges for genetic diseases

- Lack of treatment options, 95% no treatment
- Diagnosis delays and misdiagnosis 5-7 years
- 30% of patients will not see their 5th birthday

AJMC

Not So Rare: 300 Million People Worldwide Affected by Rare Diseases

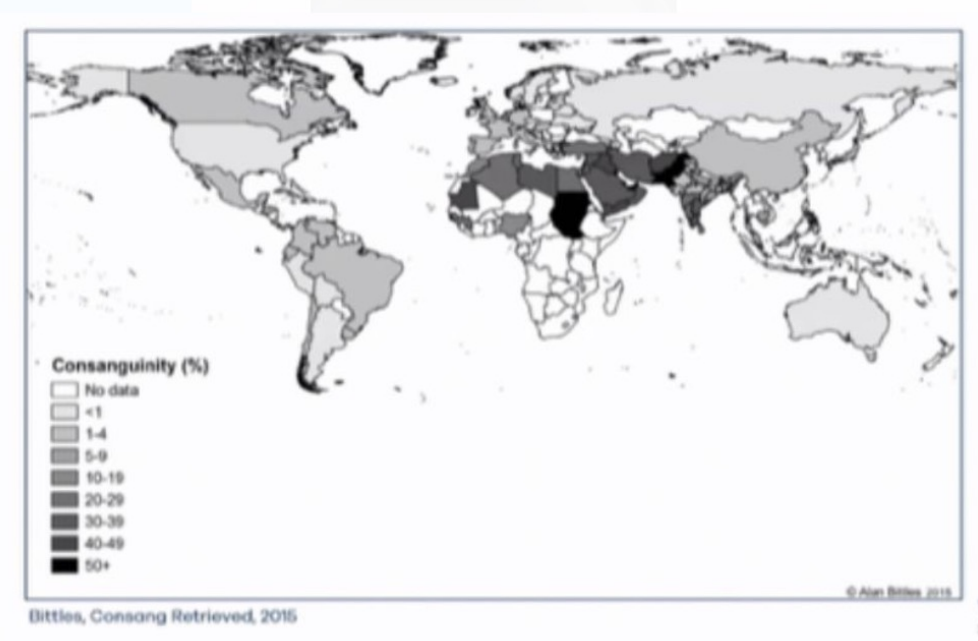
Why Rare Diseases Are Really Not So Rare

Despite recent advances in treating rare diseases, patients often feel isolated, with a proper diagnosis sometimes hard to come by.

Rare Genetic Diseases – Middle East perspective

- Middle East community has a **unique tribal structure** that is still preserved over generations. This structure **creates a sort of genetic isolates** that can be significantly used in linking medical conditions with the tribes

- Rapid rate of **growth**, **large family size**, **consanguineous marriages**, and the presence of **genetic isolates** have **greatly impacted** the **landscape of genetic disorders** in the Middle East.



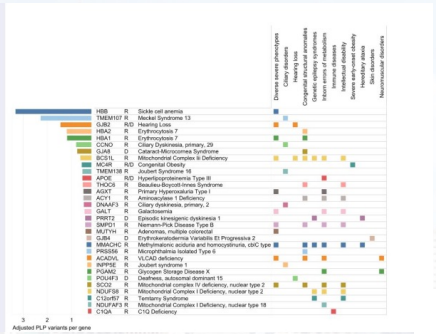
Bittles, Consang Retrieved, 2015

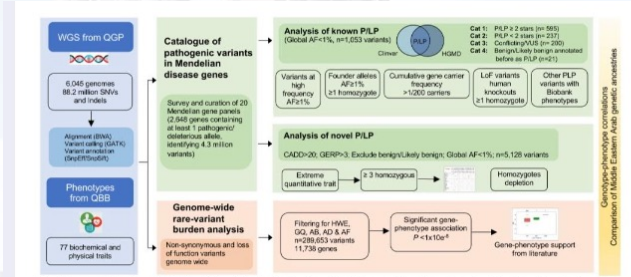
About 20% of the world population prefers consanguineous marriages (Tadmouri et al, 2009) because of their social benefits

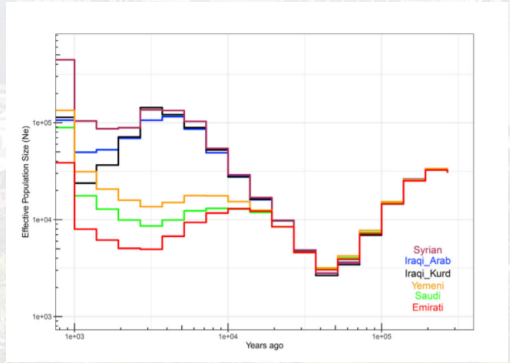
Rare Genetic Diseases – Middle East Perspective

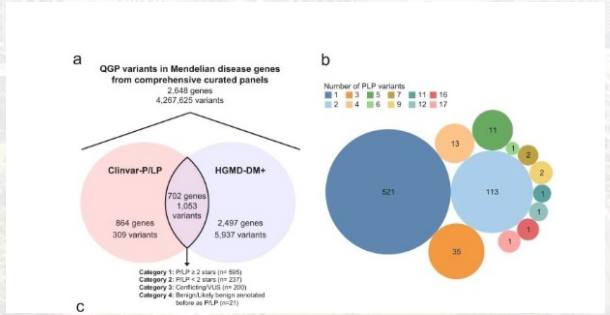
- **Gulf region has an increased rate of inherited genetic diseases—nearly double the rate in Europe and the United States**.

- Often the diseases have **never been seen before** and many disorders have **been first described** in Middle Eastern patients.









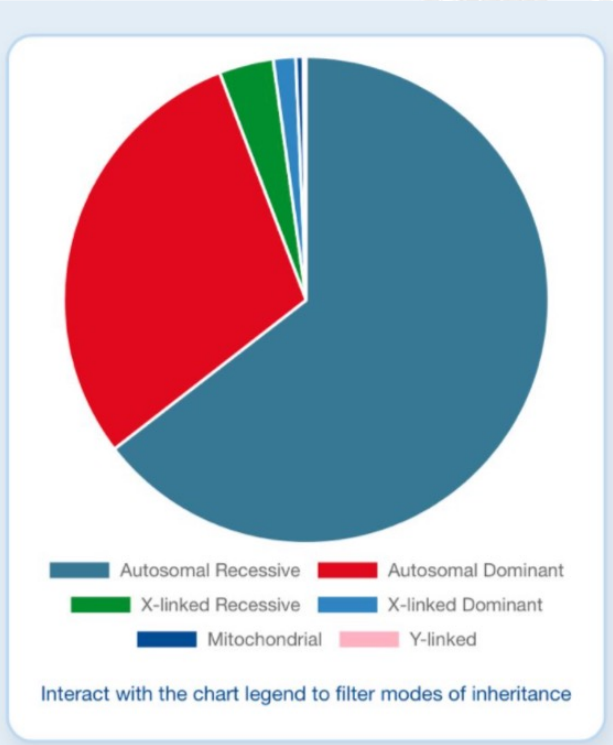
Rare Genetic Diseases – Middle East Perspective

Higher prevalence of recessive genetic disorders
Higher birth defects
Increased homozygosity

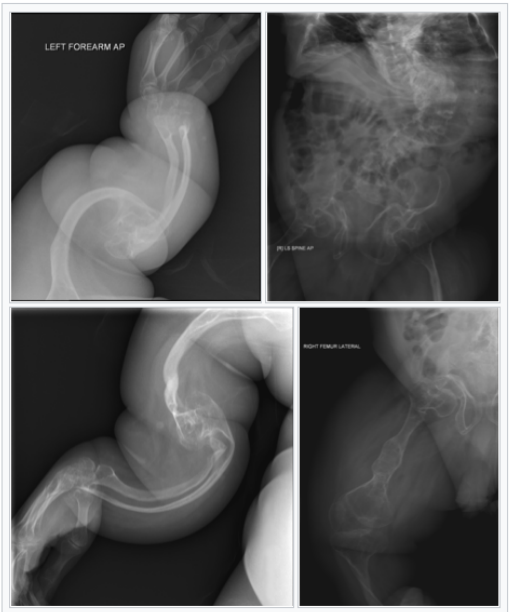
Country	Birth defects per 1,000 live births	First-cousin marriage rate in %
Sudan	82.0	46.7
Saudi Arabia	81.3	33.5
Benin	77.9	n.a.
Burkina Faso	77.0	n.a.
Occupied Palestenian Territory	76.6	n.a.
UAE	75.9	25.2
Tajikistan	75.2	n.a.
Iraq	75.2	32.3
Kuwait	74.9	24.3
Afghanistan	74.9	n.a.
USA	47.8	n.a.
Germany	43.8	n.a.

The **diagnostic strategies should be tailored to the needs of local population** and be **compatible with the cultural traditions** and social make up and the legal system of the region.

Example: Osteogenesis Imperfecta
Caucasian: Dominant
Middle Eastern: Recessive

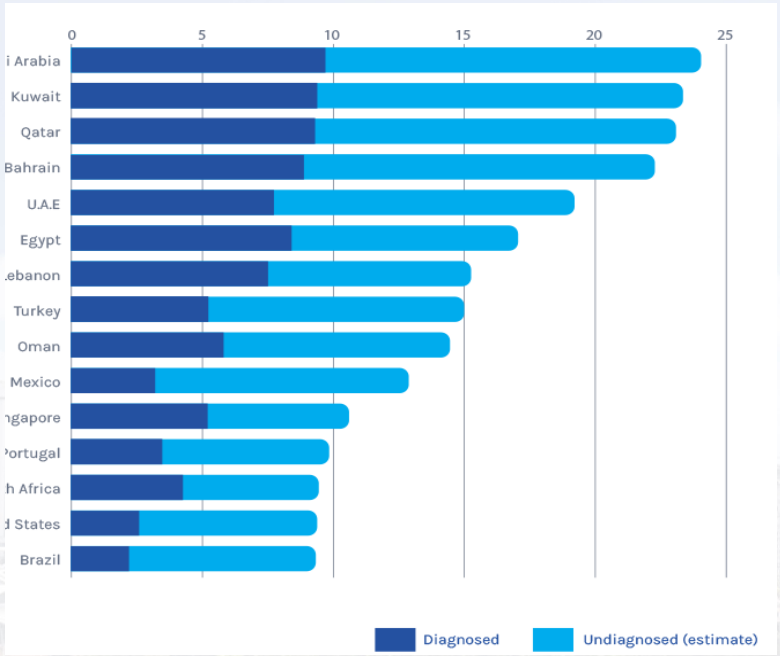


DISTRIBUTION OF THE MODES OF INHERITANCE OF GENETIC DISORDERS IN THE ARAB WORLD



Non-Communicable Diseases – Middle East perspective

According to the World Health Organization (**WHO**), the global population aged 60 years and above is expected to **double by 2050**, and **triple by 2100**.



KEY POINTS

- **Cardiovascular disease**, **cancer** and **obesity** represent up to **one third of disease burden in the MENA region**.
- In Saudi Arabia, **NCD** prevalence is **32,15% responsible for 73% of all deaths**.
- **Obesity** prevalence **41% in males** and **78% in females**.
- **Diabetes** prevalence **18,3%**.
- Our last 10 years is lived in poor health poor span

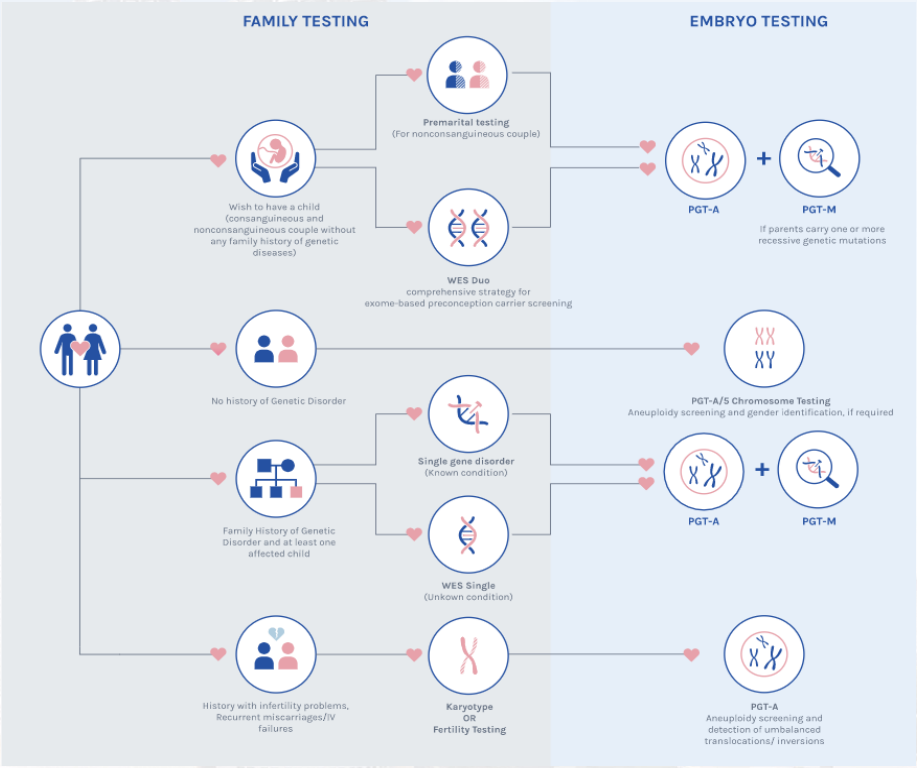
High prevalence of Diabetes, Cardiovascular and metabolic diseases. 9.4% of the population in US has Diabetes. The situation in the Middle East is almost double this number.

Infertility – Middle East perspective

Key Factors

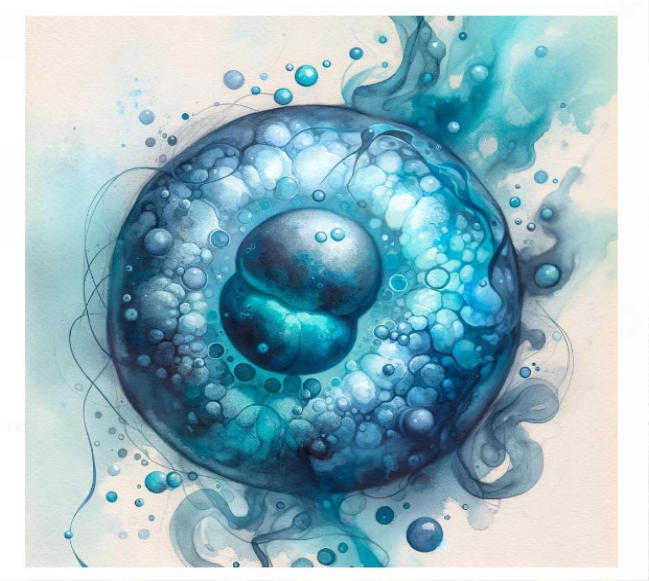
- Obesity, Smoking, Longer reproductive cycle
- Higher maternal age
- Low Fertility rates in the population (3.2 in early 2000's to 1.7-2.3 in 2015)
- Social pressures to have a baby
- Consanguinity - about 50% or higher in the GCC
- **Local factors. The association between lower fertility, parental consanguinity, obesity, and vitamin D deficiency was already described**.

Infertility is worldwide acknowledged as a major health problem. The prevalence of infertility in women of reproductive age has been estimated to be **one in every seven couples** in the **western world** and **one in every four couples in developing countries**



Health Economics – Middle East Perspective

RISING HEALTHCARE COST AND CHRONIC DISEASES PUT CONSTANT PRESSURE ON PAYERS WITH INCREASED EXPENDITURE AND LIMITED RESOURCES:



Constrained healthcare budgets:

- Increasing population
- Competitive environment
- Increasing prevalence of chronic diseases
- Increasing cost of treatment
- Lunch of high cost innovative drugs
- Higher Prevalence of rare genetic disorders in the Middle East than in Western Countries
- Poor awareness about personalized genomics testing
- Lack of tailored screening and preventive strategies

Genomics of disease risk in globally diverse populations

Global differences in the prevalence and distribution of diseases and their risk factors are **a complex phenomenon determined** by **population history**, **adaptive evolution**, **environmental, social, demographic, cultural and genetic factors**.

Understanding global genetic diversity and its impact on human health and disease has the potential to provide additional insights into the biological mechanisms underlying disease risk

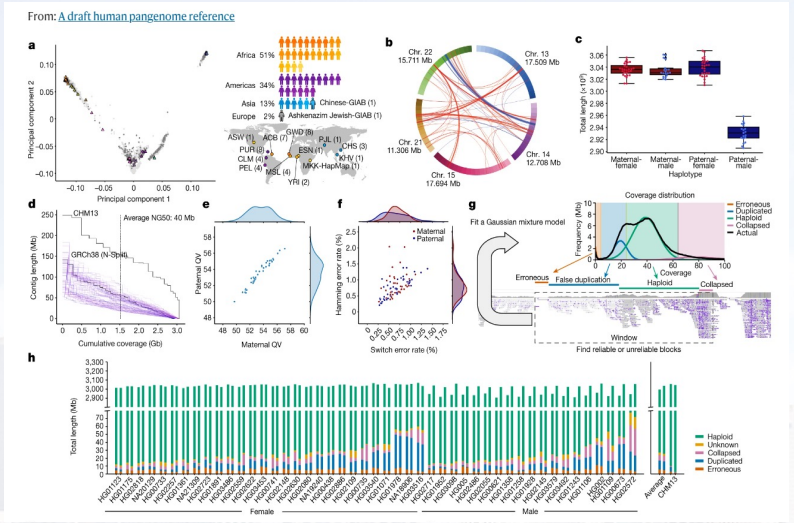
Table 1 | **Characteristics of specific populations and cohorts that facilitate genetic discovery**

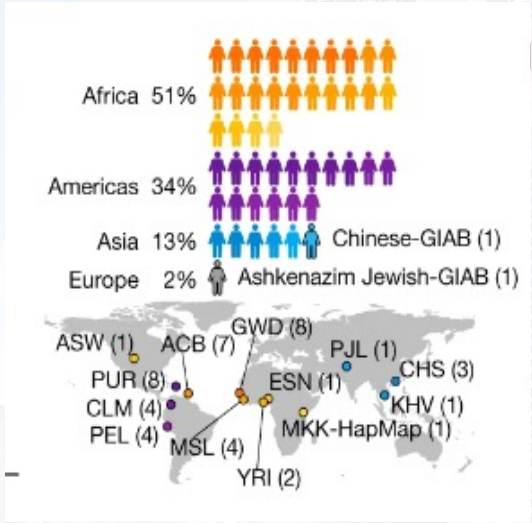
Population	Characteristic	Opportunities
Genetically diverse populations (for example, African populations)	High levels of genetic variation among individuals in the population	Novel discovery of loci associated with traits relative to less diverse populations — for example, population-specific variants, variants common in these populations but rare in other well-studied populations
Population isolates, founder populations (for example, Amish populations, Greek isolates)	Low effective population size, relative genetic homogeneity, enrichment for some rare deleterious variants	Novel discovery among loci that have increased to high frequencies in these populations but are rare in most other global populations
Populations with high levels of consanguinity (for example, Middle-Eastern populations)	High levels of homozygosity	Assessment of pathogenic potential of rare variants in homozygous form and gene function by assessment of naturally occurring gene knockouts
Admixed populations (for example, African-Americans)	Genomes of individuals are a mosaic of haplotypes of different ancestral origin	Assessment of the association between local ancestry with disease (where disease susceptibility is known to vary among source populations). Cases with disease will be enriched for specific ancestry at loci associated with disease
Populations exposed to different environmental stimuli (for example, sub-Saharan African populations exposed to malaria)	Genetic adaptation in response to environment stimulus	Adaptation, including selective sweeps or balancing selection leading to certain alleles rare or absent in other populations reaching higher frequencies in these populations (for example, the sickle cell variant associated with malaria)
Multi-ethnic cohorts	High levels of differentiation between different ethnic groups studied and different linkage disequilibrium patterns	Better resolution of causal variants associated with traits or diseases
Family-based cohorts	Pedigrees with related individuals (diseased and healthy), with detailed phenotyping for each pedigree	• Assessment of loci associated with Mendelian disease; discovery of de novo mutations associated with disease • Assessment of heritability of complex traits, accounting for shared environment

Gurdasani, D., Barroso, I., Zeggini, E. *et al.* Genomics of disease risk in globally diverse populations. *Nat Rev Genet* **20**, 520–535 (2019).

Lack of proper representation of Arab population in the reference genome

The **current reference human genome** is **nearly 20 years old** and **mostly derived from a single Caucasian individual.**) It suffers from a **major limitation**: as cannot represent the diversity found in human genomes globally in a single reference sequence.

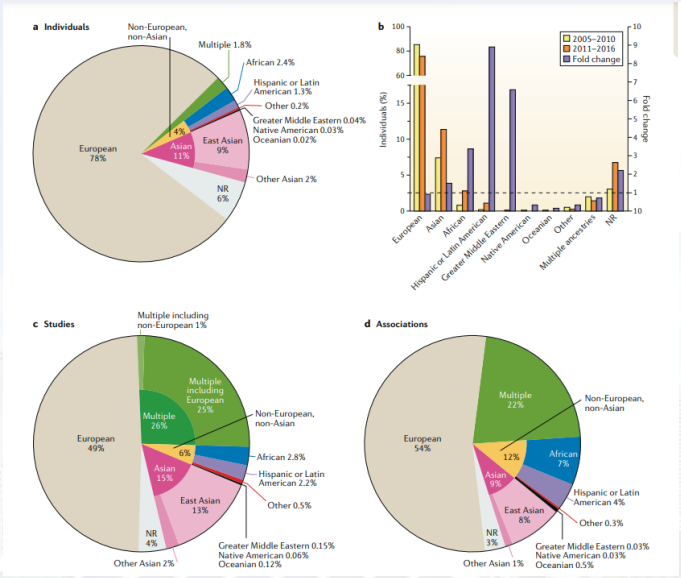


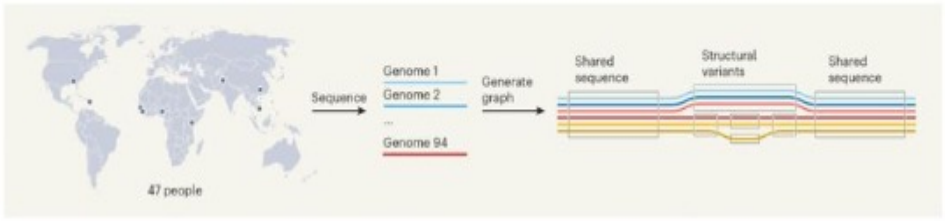


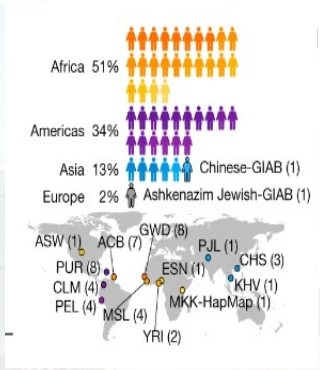
The human pangenome based on 47 individuals representing a wider global genetic diversity has been just published by the Human Pangenome Reference Consortium (HPRC).

Lack of proper representation of Arab population in the reference genome

Unfortunately, **Arab population** is still **underrepresented in the human reference genome**. As a consequence, many of Arab patients are receiving **false positive/negative or inconclusive** reports.

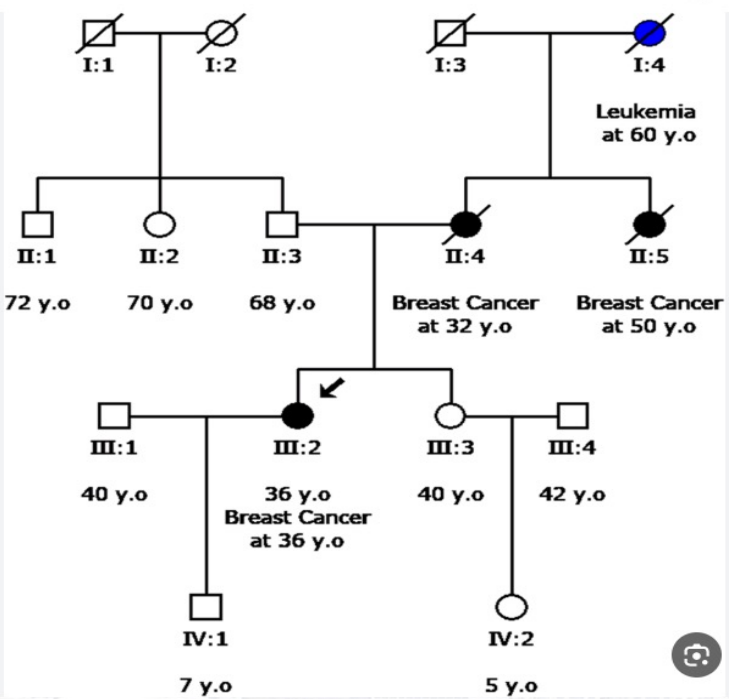






Lack of proper representation of Arab population in the reference genome

Pedigree of a family with strong breast cancer history



~35-67% of cases with GUS or VUS

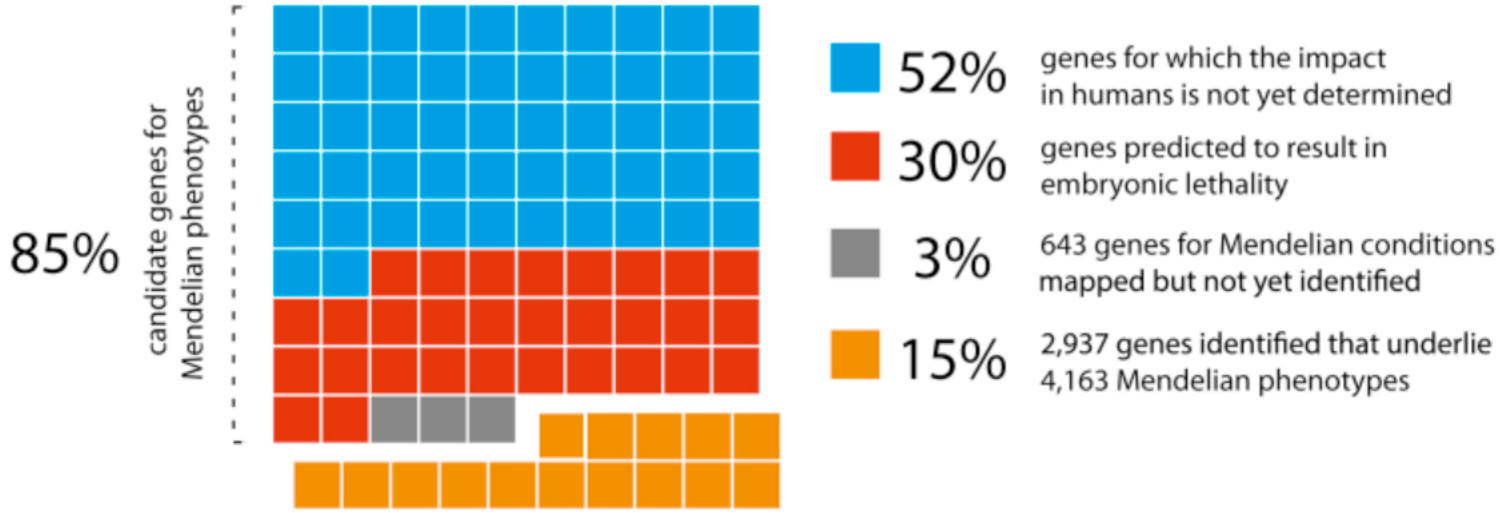
Positive family history of the disease, but there is **NO clear pattern that is consistent with any of the known inherited syndromes**

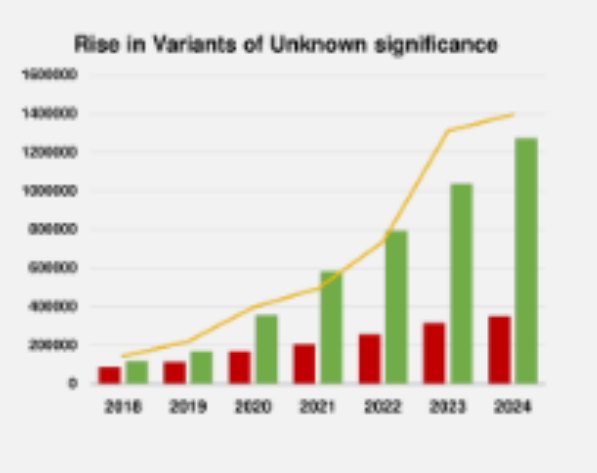
How to solve this dilemma?

- **Segregation studies**
- **Clinical Exomes**
- **WGS – National Genome Projects** (to be able to identify more genomic variants from various populations that can provide insights into health and disease)
- **Functional Studies**

Lack of proper representation of Arab population in the reference genome - GUS and VUS

Humans have approximate 20,000 genes





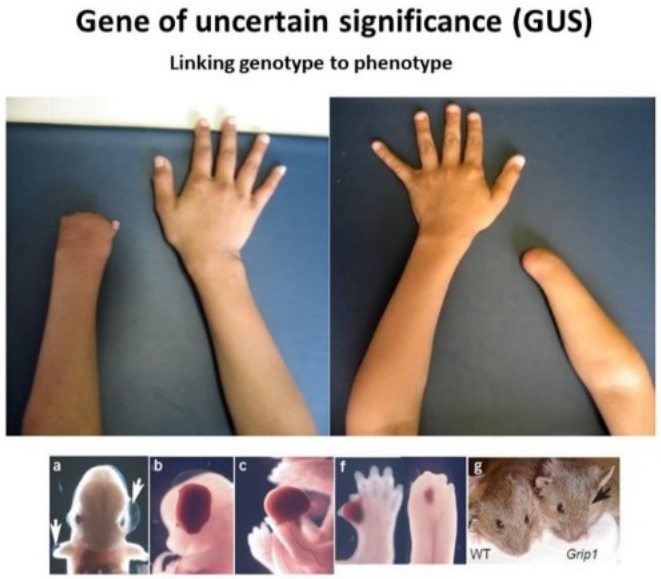
More than 50% of genes without any known function (GUS)!

VUS constitute ca. **48% of over 2 million variants** in ClinVar and **30-35%** of genetic reports remain inconclusive due to a VUS being the primary finding. Those numbers are much higher for **underrepresented ethnic groups**.

Segregation and **functional studies** can provide crucial insights to help clarify the pathogenicity of the variant and it's impact in human

Lack of proper representation of Arab population in the reference genome - GUS and VUS

Relationship between **Human Protein Coding Genes** and **Mendelian Phenotypes** requires constant research and development . While treatment decisions should **not be made based on GUS or VUS**, the ACMG recommends follow-up testing, potentially leading to reclassification that could be a key to unlocking a patient diagnosis.



~**19,000** protein-coding **genes** are predicted to exist in the human genome. Variants that cause **Mendelian phenotypes have been identified** in ~2,937 genes (~**15.5%**). Genes underlying ~643 Mendelian phenotypes (~3.38%) have been mapped but not yet identified. On the basis of analysis of knockout mouse models, LOF variants in up to ~30% of genes (~5,960) could result in embryonic lethality in humans.

For a minimum of **~52% of genes (~10,330),** the impact in humans **has not yet been determined.** Collectively**, ~16,063 genes remain candidates for Mendelian phenotypes.**

Discovering the genetic basis of a Mendelian phenotype establishes a link between genotype and phenotype**, making possible carrier** and **population screening and direct diagnosis.**

Such discoveries also contribute to **our knowledge of gene function**, **gene regulation, development, and biological mechanisms** that can be used for **developing new therapeutics**.

Importance of the genomic initiatives in the Middle East

National genomes programs **are critical** as they will **fill this gap** and **provide a road map of the reference human genome** and a **landscape of mutations in genomes** across many disease types in the Middle Eastern patients.

- This will allow us to classify properly genetic variants. Reduce number of VUS and GUS
- To understand better the molecular mechanism of diseases and the rationale for molecule-guided therapies.
- It will help us to adjust current screening and preventive strategies for the Middle Eastern populations.

Combined in-depth **genetic analysis** with **rich biobank phenotypes** to uncover **potentially 'novel' disease genes**.

We believe this approach will become more common across large-scale biobanks in the future and will have implications for **precision health** and **personalized medicine**

Importance of the genomic initiatives in the Middle East

RESEARCH Open Access

Burden of Mendelian disorders in a large Middle Eastern biobank

Waleed Aamer[1†], Aljazi Al-Maraghi[1†], Najeeb Syed[2], Geethanjali Devadoss Gandhi[1], Elbay Aliyev[1], Alya A. Al-Kurbi[1], Omayma Al-Saei[1], Muhammad Kohailan[1], Navaneethakrishnan Krishnamoorthy[1], Sasirekha Palaniswamy[1], Khulod Al-Malki[1], Saleha Abbasi[1], Nourhen Agrebi[1], Fatemeh Abbaszadeh[3], Ammira S. Al-Shabeeb Akil[1], Ramin Badii[3], Tawfeg Ben-Omran[4,5,6], Bernice Lo[1,7], The Qatar Genome Program Research Consortium, Younes Mokrab[1,8,9*] and Khalid A. Fakhro[1,7,8*]

Recent research, utilizing WGS of >6000 Qatari individuals:

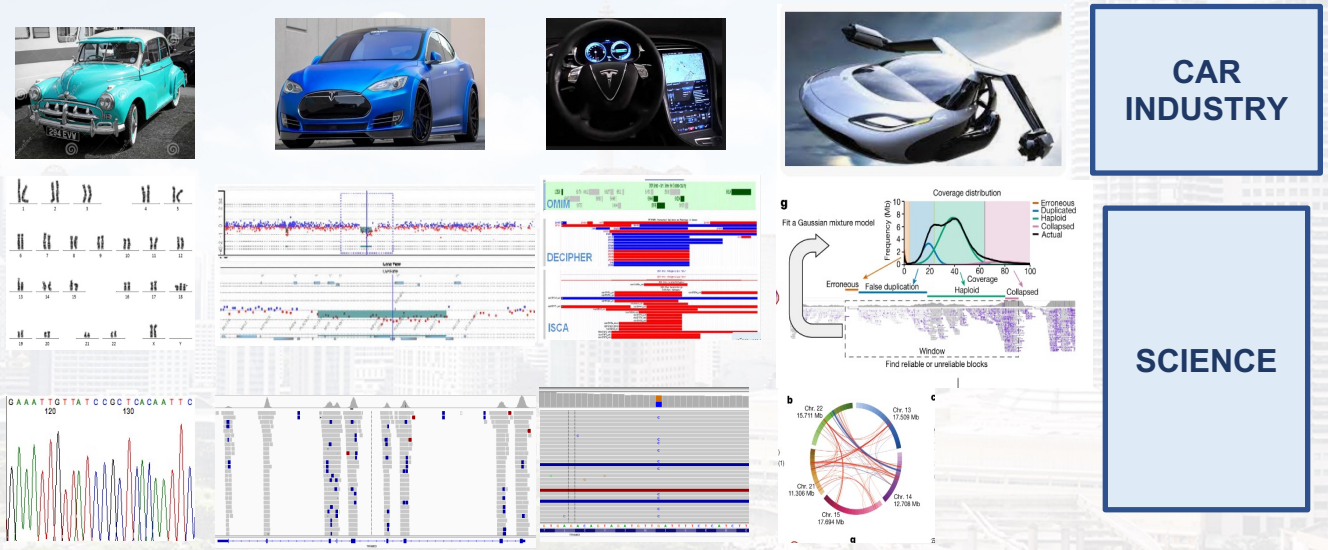
- **62.5%** of individuals carry at least **1 known disease mutation linked to recessive conditions.**
- There are **56 rare diseases** which each have a cumulative **gene-carrier frequency** (GCF) of >1/50, exceeding **the threshold** of what should be screened for in **newborn and premarital screening programs** in Qatar.
- **Discovered new genes with** high mutational load, including **5 genes with impact on traits relevant to metabolic disorder** and **type 2 diabetes**, consistent with the high prevalence of these conditions in the region.
- **Several mutations that are marked in databases as "pathogenic"** which appear in homozygous state(!) in seemingly **healthy individuals in the Qatar Biobank**, prompting their **reconsideration as potentially not really disease-causing**.

Evolvement of the genomics and personalized medicine

Recent technological advances in NGS:

- significantly **improved the diagnostic yield** both in rare **genetic disorders** for which **disease genes were unknown** and/or **genetic testing was not offered**

- provided completely new, innovative - **cell free DNA** or **single cell - diagnostic applications** such as NIPT, PGT-A/PGT-M or liquid biopsy testing



FASTER, SMARTER, MORE PRECISE, MORE EFFICIENT

60's 80's 90's 21st century

Preimplantation Genetic Diagnosis (PGT-A/PGT-M testing) as Disease Prevention Approach

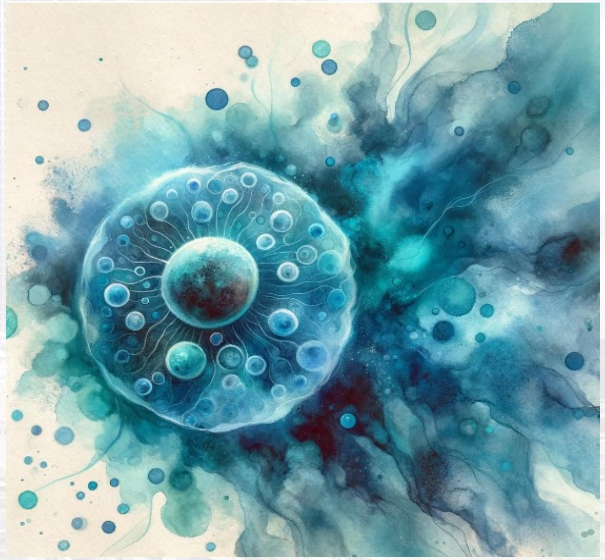
For whom?

Couples with **fertility problems**, **recurrent miscarriages**, **unexplained IVF failures** and **affected child** or with a **known family history of a particular genetic diseases.**

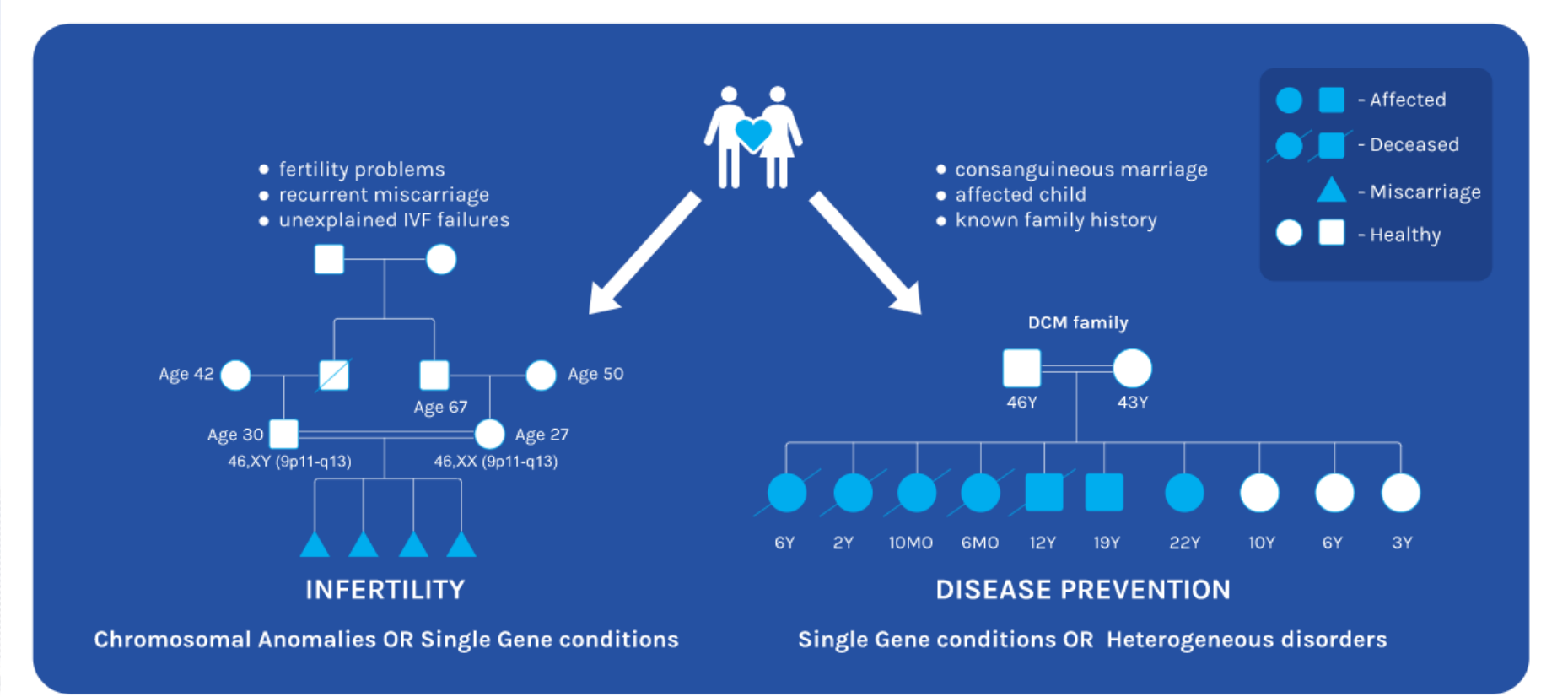
With the recent advancements in the **use of PGT-M is becoming a significant alternative for many families at risk for whom prenatal diagnosis and termination of pregnancy** of an affected fetus **is not a religiously, traditionally or culturally acceptable** option.

AIM

Promoting Reproductive Genetics as a part of Preventive strategy for the local population in order to reduce/remove genetic disorders from the population



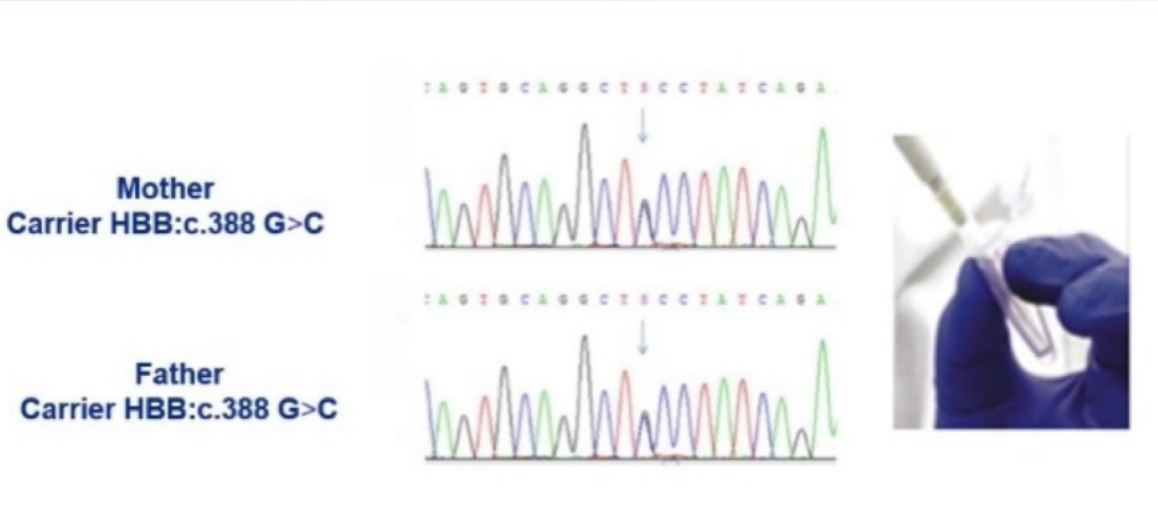
Preimplantation Genetic Diagnosis (PGT-A/PGT-M testing) as Disease Prevention Approach



Step 1: Genetic consultation
Step 2: Family testing for rare genetic disorders
Step 3: PGT-M + PGT-A + linkage testing

Preimplantation Genetic Diagnosis (PGT-A/PGT-M testing) as Disease Prevention Approach

The researches **compared** the **cost of the PGT-M intervention** versus the **cost of care for patients with most common single gene defect disease**: sickle cell disease.



They calculated that an average lifetime cost could reach as high as $1.7 million.

The only cure is **bone marrow transplant**, known as hematopoietic stem cell transplantation, with cost: between **$150,000** and **$250,000** per patient.

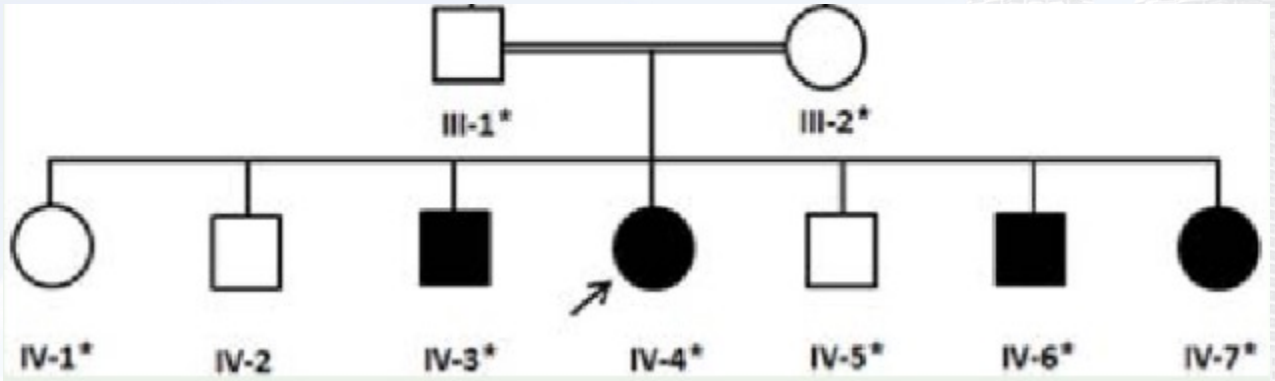
Even more promising **gene therapies** to treat sickle cell disease are expected to have a price tag of more than **$1 million**.

On the other hand, the estimated cost **of PGT-M-IVF** to prevent the dieses is **$15,000 to $25,000**.

So, PGT-M-IVF might improve overall patients' management and reduce the cost paid by local governments for the treatment of inherited diseases.

Preimplantation Genetic Diagnosis (PGT-A/PGT-M testing) as Disease Prevention Approach

Family with history of pediatric inherited metabolic disease, characterized by early-onset **diabetes**, **skeletal dysplasia** and **hepatic dysfunction**



- consanguineous marriage
- patient's mean age was 4.6 years (range: 10 months to 17.5 years)
- there was a variation in the phenotype between siblings
- diabetes was the presenting feature in all affected children
- 3 out of 4 children had severe hepatic dysfunction, followed by **LIVER TRANSPLANTATION**

Preimplantation Genetic Diagnosis (PGTA/PGTM testing) as Disease Prevention Approach

Reproductive genetics IVF + PGTM – health management per couple
IVF cycle – 20 000 AED / per couple
IVF related genetic testing – 18 000 AED /couple
An avoidable costs of medical care including unnecessary diagnostic procedures and life-long monthly treatment

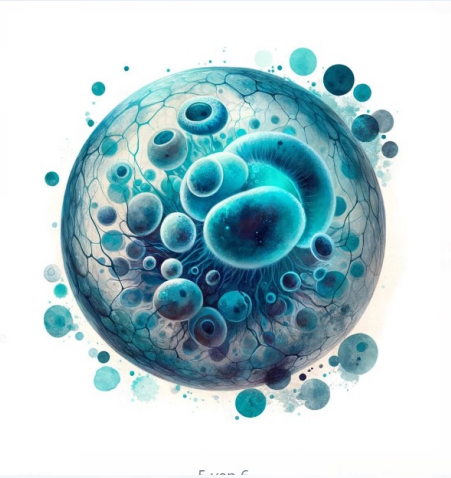
Total cost 40 000 AED (11 000 USD)

Lifetime – health management of hepatic dysfunction per patient
Liver transplant – 286 178 AED / surgery
Lifetime Immunosuppressant's treatment – 9182 AED / month
Special diet – 1430 AED / month
Uncertainty for families
Emotional drain of dealing with serve genetic disorder

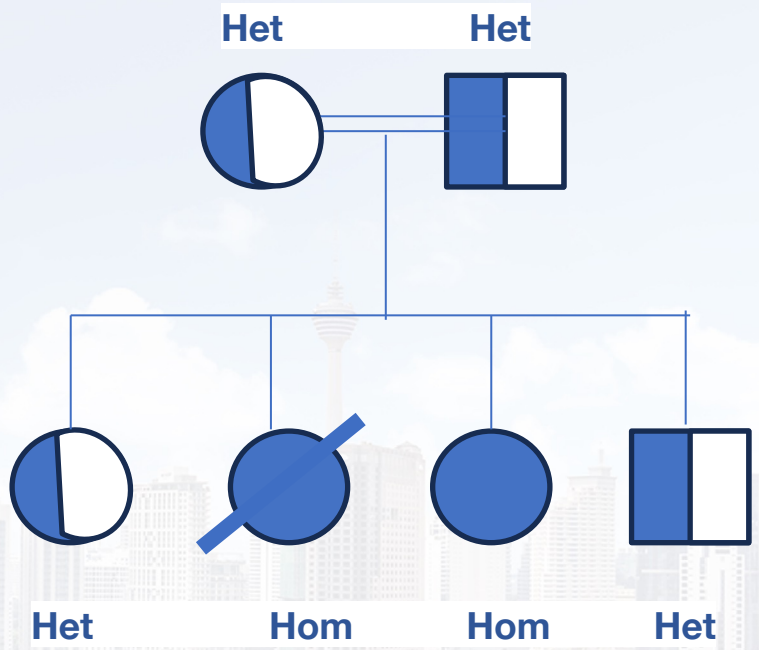
Total cost 296 000 AED (81 000 USD)

Disease prevention = Cost savings

Preimplantation Genetic Diagnosis (PGT-A/PGT-M testing) as Disease Prevention Approach



CMMRD case (MSH6 gene, c.2927G>A; p.R976H)



It's a common condition in Middle East and cause up to 30% of childhood cancers in our region.

Family with history of Constitutional mismatch repair deficiency (CMMRD) syndrome, they need to be on surveillance because of high risk of different Cancer development. One girl already died at age of 2, because of developed glioblastoma.

Preimplantation Genetic Diagnosis (PGTA/PGTM testing) as Disease Prevention Approach

Comprehensive multi OMICS solution for Sickle cell/Thalassemia

Genomics

1. CBC blood report

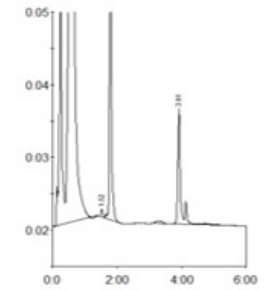
	Mean	SD	Range
RBC 10^{12}/L	5.8	0.43	5.0–6.9
Hb g/dL	12.6	1.21	10.0–14.8
MCV fL	63.8	3.74	57.2–72.5
MCH pg	21.5	1.3	19.3–24.5
MCHC g/dL	33.7	0.92	32.3–36.3
% reticulocyte	1.6	0.56	0.1–3.19
% microcytic	36.5	13.6	10–62.6
% hypochromic	3.4	2.87	0.3–12.5
%Micro / %Hypo	15.1	9.97	4.3–54
Iron µmol/L	18.1	3.95	14.8–23.1
Transf g/L	2.48	0.28	1.73–2.98
Ferritin µg/L	137	2.5	55–227
% Sat	30	5.6	22–38

People with thalassemia have fewer healthy red blood cells and less hemoglobin than normal

2. Hemoglobin electrophoresis

Patient report

Bio-Rad	DATE: 05/29/2023
D-10	TIME: 04:49 PM
S/N: #D30F513096	Software version: 4.30-2
Sample ID:	1826123ED
Injection date	05/29/2023 02:22 PM
Injection #: 13	Method: HbA2/F
Rack #: ---	Rack position: 2



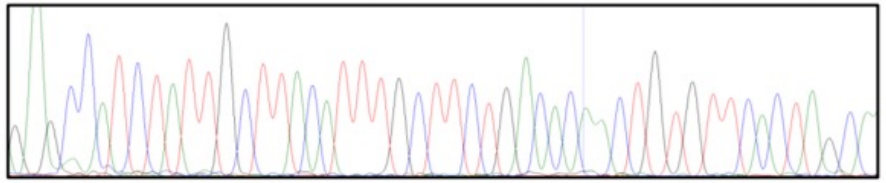
Peak table - ID: 1826123ED

Peak	R.time	Height	Area	Area %
A1b	0.26	40119	196234	13.7
LA1c/CHb-1	0.63	87286	932988	65.3
P3	1.52	1130	5212	0.4
A0	1.78	58432	215652	15.1
Unknown	3.91	15930	78391	5.5
Total Area:		1428476		

Concentration:	%
---	0.0

Clinical Pathology

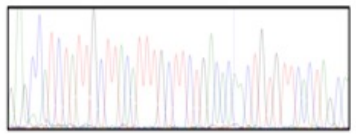
3. Mutation identification by DNA sequencing on the family members



4. PGT-M reproductive genetic testing on biopsied embryos as a disease prevention program

Embryo biopsy

PGT-A



PGT-M

24 Chromosome PGT-A by NGS and PGT-M Single Gene Disorder with HLA Matching

TEST INTERPRETATION

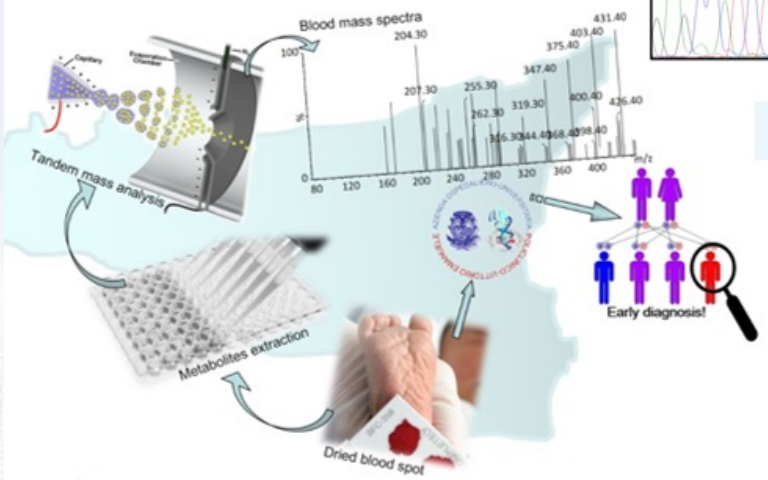
Embryo ID	PGT-A (Aneuploidy Screen)	PGT-M (Genetic diagnosis)	HLA matching	MtDNA to Autosomal score	Interpretation
SS1	**Euploid; XX**	**Carrier**	**Not Matching**	**0.8, indicative of healthier mitochondria**	**Normal Female; one mutant allele and one normal allele present for HBB: 619bp del; Suitable for transfer**
SS2	Aneuploid; Complex Abnormal	Untested	Untested		Abnormal; Not suitable for transfer
SS3	Euploid; XX	Affected	Untested		Normal Female; two mutant alleles present for HBB: 619bp del; Not suitable for transfer
SS4	Aneuploid; Trisomy 3, XY	Untested	Untested		Abnormal Male; Not suitable for transfer
SS5	Euploid; XY	Affected	Untested		Normal Male; two mutant alleles present for HBB: 619bp del; Not suitable for transfer
SS6	Aneuploid; Partial duplication of chromosome 17, XX	Untested	Untested		Abnormal Female; Not suitable for transfer
SS7	**Euploid; XY**	**Non-Carrier**	**Not Matching**	**1.0, indicative of healthier mitochondria**	**Normal Male; two normal alleles present for HBB: 619bp del; Suitable for transfer**
SS8	**Euploid; XY**	**Carrier**	**Not Matching**	**0.5, indicative of healthier mitochondria**	**Normal Male; one mutant allele and one normal allele present for HBB: 619bp del; Suitable for transfer**
SS9	Euploid; XY	Affected	Untested		Normal Male; two mutant alleles present for HBB: 619bp del; Not suitable for transfer
SS10	Aneuploid; Trisomy 22, XX	Untested	Untested		Abnormal Female; Not suitable for transfer
SS11	**Euploid; XX**	**Carrier**	**Matching**	**0.3, indicative of healthier mitochondria**	**Normal Female; one mutant allele and one normal allele present for HBB: 619bp del; Suitable for transfer**

Ensuring family that embryos will not carry Thalassemia mutation

Preimplantation Genetic Diagnosis (PGTA/PGTM testing) as Disease Prevention Approach

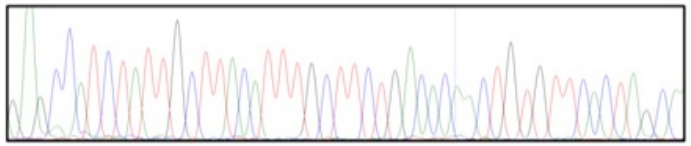
Comprehensive multi OMICS solution for new born screening

1. ELISA + Mass Spectrometry



Clinical Pathology

2. Mutation identification by DNA sequencing on the new born baby



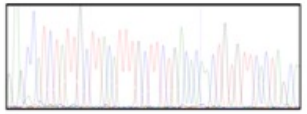
Genomics

Precision Medicine

3. Targeted therapy or gene therapy to rescue the new born baby (example SMA gene therapy)

4. For the future reproductive family planning - PGT-M reproductive genetic testing on biopsied embryos as a disease prevention program

Embryo biopsy



PGT-A +PGT-M testing

24 Chromosome PGT-A by NGS and PGT-M Single Gene Disorder with HLA Matching

TEST INTERPRETATION

Embryo ID	PGT-A (Aneuploidy Screen)	PGT-M (Genetic diagnosis)	HLA matching	MtDNA to Autosomal score	Interpretation
S51	Euploid; XX	Carrier	Not Matching	0.8, indicative of healthier mitochondria	Normal Female; one mutant allele and one normal allele present for HBB: 619bp del; Suitable for transfer
S52	Aneuploid; Complex Abnormal	Untested	Untested		Abnormal; Not suitable for transfer
S53	Euploid; XX	Affected	Untested		Normal Female; two mutant alleles present for HBB: 619bp del; Not suitable for transfer
S54	Aneuploid; Trisomy 3, XY	Untested	Untested		Abnormal Male; Not suitable for transfer
S55	Euploid; XY	Affected	Untested		Normal Male; two mutant alleles present for HBB: 619bp del; Not suitable for transfer
S56	Aneuploid; Partial duplication of chromosome 17, XX	Untested	Untested		Abnormal Female; Not suitable for transfer
S57	Euploid; XY	Non-Carrier	Not Matching	1.0, indicative of healthier mitochondria	Normal Male; two normal alleles present for HBB: 619bp del; Suitable for transfer
S58	Euploid; XY	Carrier	Not Matching	0.5, indicative of healthier mitochondria	Normal Male; one mutant allele and one normal allele present for HBB: 619bp del; Suitable for transfer
S59	Euploid; XY	Affected	Untested		Normal Male; two mutant alleles present for HBB: 619bp del; Not suitable for transfer
S510	Aneuploid; Trisomy 22, XX	Untested	Untested		Abnormal Female; Not suitable for transfer
S511	Euploid; XX	Carrier	Matching	0.5, indicative of healthier mitochondria	Normal Female; one mutant allele and one normal allele present for HBB: 619bp del; Suitable for transfer

Ensuring family that embryos will not carry SMA mutation

Summary:

- Prevalence of rare genetic disorders is much higher in the Middle East than in Western Countries.
- Testing Arab samples in the European or US Laboratories might generate FALSE negative or False Positive results, not mention about data privacy and security.
- Preventive programs must be tailored to the genetic makeup of the local population.
- Genome sequencing of large biobanks from under-represented ancestries provides a valuable information about Mendelian diseases.
- Promote Reproductive Genetics as a part of Preventive strategy in order to reduce/remove genetic disorders from the population with the high rates of consanguinity.

21ST PGDIS CONFERENCE

PGDIS

21ST PGDIS CONFERENCE

PGDIS

THANK YOU

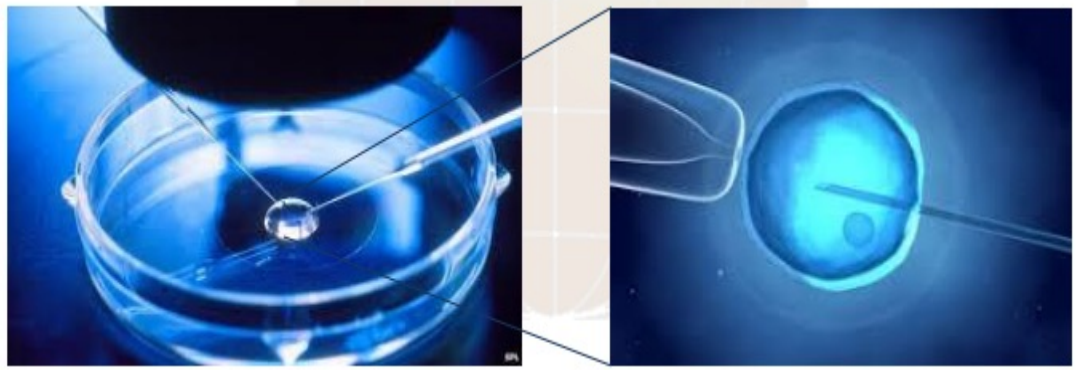
PGT and BEYOND...

Evolvement of the genomics and personalized medicine

The availability of high advanced PGT-M genetic services locally will not only provide accurate genetic diagnosis, but also might be offered as a disease prevention program.

It will avoid an uncertainty for families, not to mention the economic costs of unnecessary diagnostic procedures.

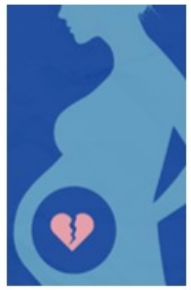
It will improve patient's management, survival rate and reduce the spent amount and cost paid by local governments for the treatment of chronic diseases for local community.



Preimplantation Genetic Testing for Monogenic Disorders (PGT-M) as an inherited genetic disease prevention program.

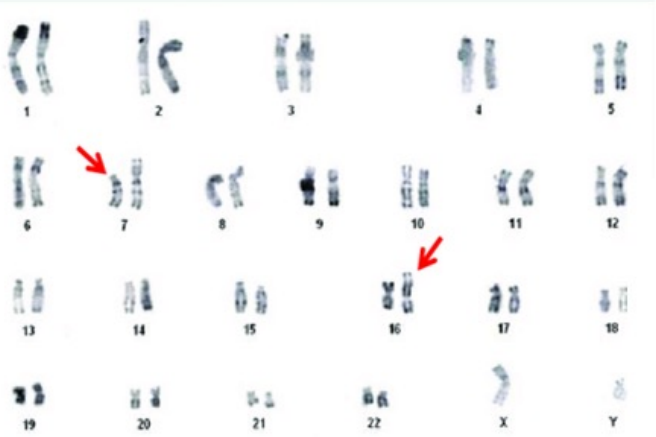
Preimplantation Genetic Diagnosis (PGTA/PGTM testing) as Disease Prevention Approach

Improving reproductive health by comprehensive multi OMICS solution



Recurrent miscarriages
or
IVF failures
(approximately 25% of recognized pregnancies end in miscarriage)

Classical Cytogenetics



Karyotype of a patient with a balanced reciprocal translocation:
46, XY, t(7;16) (p11;p13).

Genomics

PGT-A Reproductive Genetic testing to make sure that baby will be without any chromosomal anomalies