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GPDx

Genomic Precision Diagnostic

by Igenomix®

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FACTS

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THE DIAGNOSTIC DISEASES ODYSSEY

Too many families are bounced around from physician to specialist and back again only to receive multiple genetic misdiagnoses.

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40% of rare disease patients are misdiagnosed at least once.

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2-3

Incorrect diagnoses are provided wrongly for a given condition.

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5 YEARS

The average time to obtain a correct diagnosis for rare diseases.

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7

Physicians to be seen by patients and families before a diagnosis is made.

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Patients and families often do not receive proper Information to help them cope with their diagnosis and condition.

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Limited knowledge of diagnostic options for doctors and updated state of the art using new technologies.

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WHY IGENOMIX CAN HELP YOU?

Igenomix®
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SUPPORTIVE PROFESSIONAL

FREE GENETIC COUNSELING

Certified Genetic Counselors to guide you and your patients choosing the right genetic testing.

Dr Bratati Chaudhary

- Ph.D in Biotechnology and Molecular Biology from Birla Institute of Technology and a Post Doctoral experience from IIIT, Hyderabad.
- More than ten (10) years of experience in genetics, cytogenetics, molecular laboratory, and genetic counseling.

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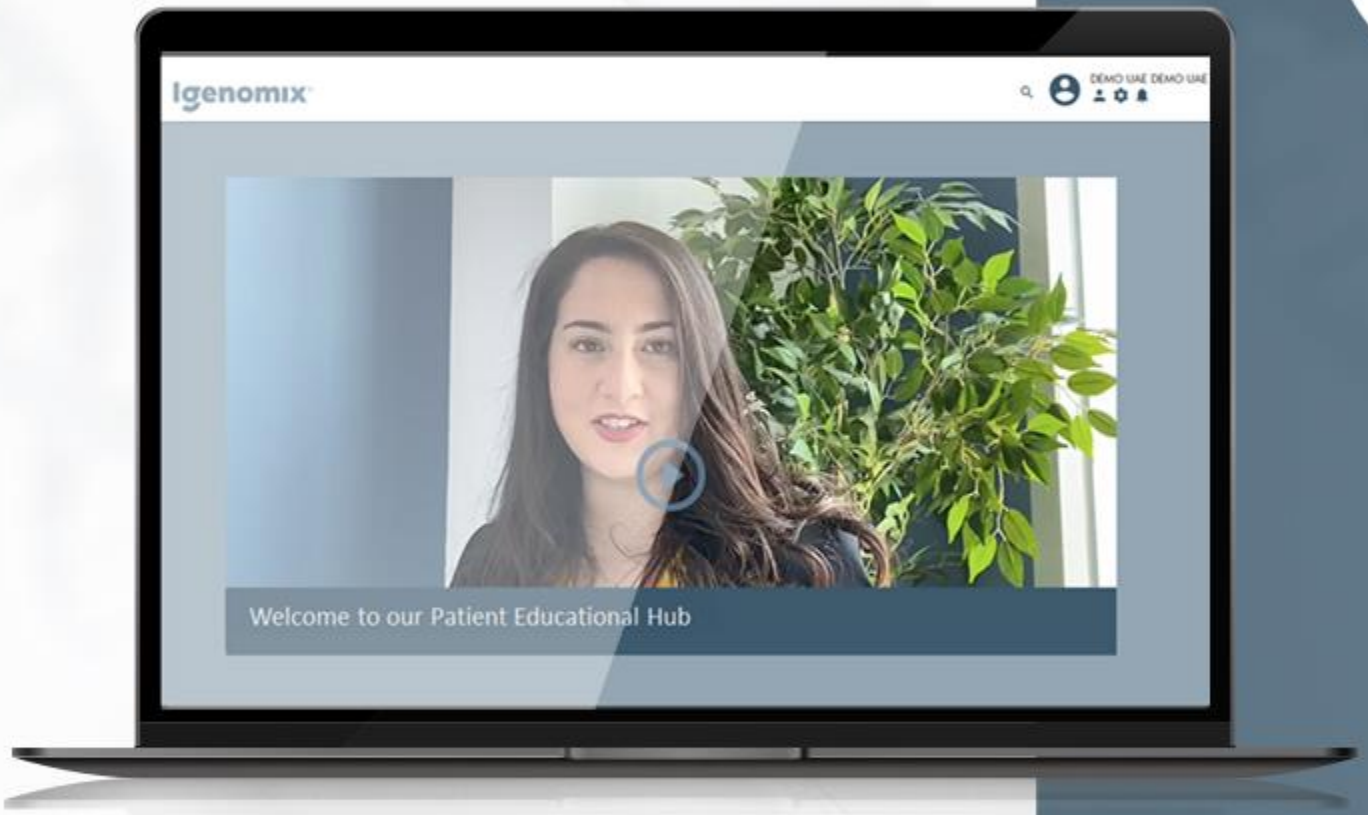

PATIENT FRIENDLY

GENETIC COUNSELING E-LEARNING PLATFORM

Use the e-learning platform to access to educational material and schedule meetings with our Genetic Counselors.

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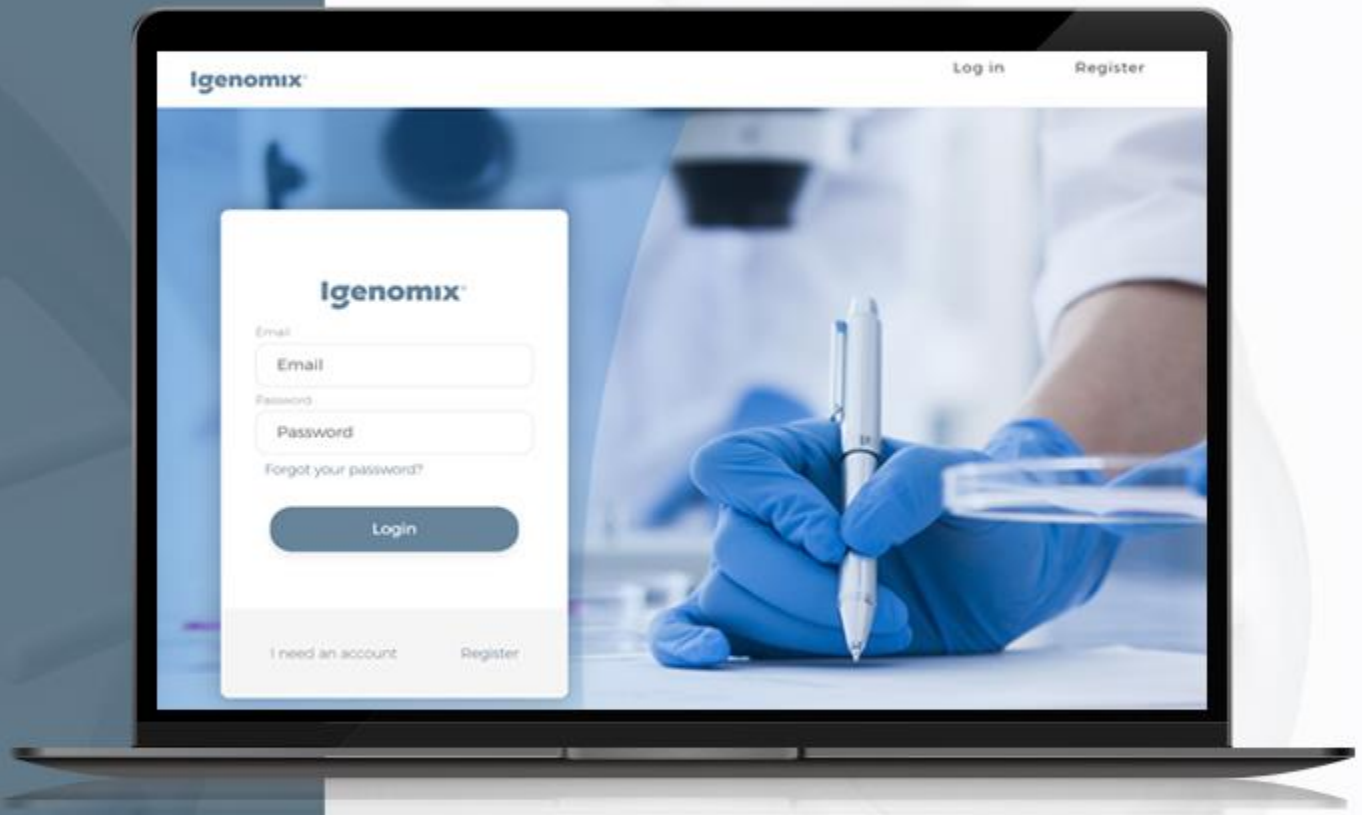
QUICK
EASY

ONLINE TEST MANAGEMENT

Use the online platform to check the tests status and download documents and reports.



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360

DEDICATED CONCIERGE

CUSTOMER SUPPORT

One contact to unify and manage everything during the testing cycle.

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All TECHNICAL CAPABILITIES IN-HOUSE

Testing, analysis, and reporting for all genetic services

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100%

IN-HOUSE SEQUENCING AND BIO IT

State-of-the Illumina platform and bioinformatics pipeline.

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TOP QUALITY SEQUENCING

- Illumina best supplier for NGS related operations
- Latest platform – NOVASEQ 6000
- >100x exome mean coverage
- >98% exome CDS bases covered >20x
- >98% variants in HGMD covered
- >98% variants in Clinvar covered
- Best in class CNV detection included in the report.
- Mitochondrial analysis included in reports

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ACCURATE ANALYSIS, INTERPRETATION & REPORTING

Using international and most relevant Databases

- Allele Frequency checked by human population databases (ExAC, 1000G, gnomAD, igenomix internal database)
- Genetic inheritance pattern by (OMIM)
- Variants analysis based on disease updated databases (HGMD, ClinVar, ClinGen, igenomix internal database)
- Literature search by (PubMed,Google scholar)
- In addition to SNVs and small Indels, copy number variants (CNVs) are detected using the Exome Depth method
- Pathogenicity of variants determined by American College of Medical Genetics (ACMG)
- Reports include a full clinical interpretation of results

gnomAD genome aggregation database · ClinVar · OMIM® · HGMD® · ClinGen Clinical Genome Resource · ACMG American College of Medical Genetics and Genomics · PubMed

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SKILLED EXPERIENCED PhD SCIENTIFIC COMMITTEE

High-Quality interpretation by rare disease team with vast experience in Middle East

Dr. Garcia-Planells,

PhD Human Genetics

Clinical_Director and Scientific Advisor Rare Diseases, H.Q.
With +20 years of experience. European Molecular Genetics Quality Network (EMQN) Assessor. He is the President of the Spanish PrenatXC3 al Diagnostic Association (AEDP). Member of the board of directors of the Ibero-american Society of Prenatal Diagnostic and Treatment (SIADTP)

Dr. Julio Martín

PhD Molecular Genetics

Director of Applied Clinical development and Data Curator, H.Q.
With +17 years of experience, He has been director of the Laboratory of Molecular Diagnosis, (Preimplantation Genetic Diagnosis Unit) since 2003. His main interest is in the field of genetics of single-gene disorders, Exome, and genome sequencing as well as analysis, curation, and interpretation.

Dr. Lova Satyanarayana

PhD Human Genetics

Director Genomic Precision Diagnostic, Middle East.
Having 10+ years of research experience in the field of genomics and molecular biology with 13 publications in peer reviewed national and international journals. He has a vast experience and is currently responsible for analyzing and interpreting genetic test results for (Genome-Exome, targeted panels, mitochondrial genome, Microarray, etc)

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LOCAL PROFESSIONALS HELPING YOU IN EVERY STEPS OF THE PATIENT JOURNEY

Francisco Rodriguez Herrera
Managing Director
Middle East&India

Vikram Ganju
Country Manager India

Dr. Vandana Sharma
Lab manager

Dr Bratati Chaudhary
Genetic Counselor

Rahul Kadwal
Logistics & Customer Support

Ayman Ragheb
Marketing Manager
Middle East & India

PROCESS & SAMPLE REQUIREMENTS

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IGENOMIX MANAGES & SUPPORTS YOU DURING ALL THE PROCESS

	Sample type	Container	Transportation Temperature	Volume
	Peripheral blood	EDTA vacutainer	20-25°C	3 – 4 ml
	Purified genomic DNA	In a sealed Eppendorf tube	20-25°C	A minimum 1 microgram of DNA at a concentration of 50-100ng/µl
	POC (fetal tissue)	Tissue in sterile container in saline and cardiac or cord blood in vacutainer	20-25°C	3 – 4 mm POC specimen or 50 100 mg of each tissue
	Amniotic Fluid	Sterile container	20-25°C	10-15ml
	Chorionic villi	Sterile container with culture medium or saline solution with 1% antibiotic	2-8 °C	300-500mg

IGENOMIX GENETIC TESTS PORTFOLIO

Igenomix®
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IGENOMIX PROVIDES ADVANCED GENETIC SERVICES IN TWO MAIN AREAS

Genetics for Family diseases
Diagnostics and Precision Medicine

Genetics for Treating Infertility and having a healthy Pregnancy

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GENETIC DIAGNOSTIC TESTS

Genome/Exome

- WES TRIO
- WES COUPLE
- WES INDEX

Chromosomal

- CMA — Chromosomal MicroArray

Single Gene Sequencing

- EXP — Repeat expansion analysis
- MLPA — Multiplex ligation-dependent probe amplification
- NGS — Next Generation Sequencing
- SANG — Sanger/MiniSeq sequencing

Precision Panels

- Cardiology
- Metabolic
- Endocrinology
- Gastroenterology
- Reproductive
- Neurology
- Haematology
- Dermatology
- Ophthalmology
- Rare Diseases
- Osteology
- Pneumology
- Ear, Nose Throat
- Connective Tissue Disorder
- Oncology
- Nephrology
- Immunology
- New Born Testing
  - Screening
  - ICU Diagnostic

DIAGNOSTIC TESTS APPLICATIONS

Preconception

For people who want to know if carry a genetic mutation and determine if there are at risk of having a child with a genetic disease.

Prenatal

Method of testing for common chromosomal abnormalities that can occur in a developing baby.

Neonatal

Is performed in order to identify or rule out the mutation that causes the symptoms, and in a lot of cases to confirm diagnosis.

Childhood / Adulthood

Is used when no symptoms are shown yet. But there is probability of carrying the mutation and developing symptoms later In life.

Cardiology
Cardiomyopathy gene panel
Cardiac Channelopathy gene panel

Connective Tissue Disorders
Ehlers-Danlos syndrome gene panel
Marfan syndrome gene panel
Cutis Laxa gene panel

Dermatology
Ectodermal dysplasia gene panel
Epidermolysis bullosa gene panel
Ichthyosis gene panel
Oculocutaneous albinism gene panel
Xeroderma pigmentosum gene panel
Tuberous Sclerosis (TSC1 & TSC2) gene panel
TSC1 deletion/duplication
TSC2 deletion/duplication

Endocrinology
Monogenic and syndromic obesity gene panel
Hyperlipidemia gene panel
Maturity-onset diabetes of the young (MODY) & neonatal diabetes gene panel
Disorders of Sex Development (Abnormal Genitalia) Panel
Hereditary pancreatitis gene panel
Congenital adrenal hyperplasia gene panel
Congenital adrenal hyperplasia CYP21A2 (21-0H)
Congenital adrenal hyperplasia CYP21A2 (21-0H) deletion/duplication analysis

ENT
Deafness (syndromic & non-sydromic) gene panel
Waardenburg syndrome gene panel
Usher syndrome gene panel
Bronchio-Oto-Renal syndrome panel

Gastroenterology
Alagille syndrome gene panel
Congenital hepatic fibrosis gene panel
Hyperbilirubinemia gene panel
Hemochromatosis gene panel
Progressive familial intrahepatic cholestasis gene panel
Wilson disease (ATP7B) gene analysis
ATP7B deletion/duplication

Haematology
Hereditary Hemorrhagic Telangiectasia
Congenital afibrinogenemia gene panel
Bone marrow failure syndrome gene panel
Anemia gene panel
Haemophilia (F8 & F9) gene panel
F8 intron 22 inversion
Hemophagocytic lymphohistiocytosis (HLH) gene panel
Beta-thalassemia (HBB) gene analysis
Alpha-thalassemia (HBA1/2) gene analaysis
HBA1 & HBA2 deletion/duplication
Von Willebrand disease (VWF) gene analysis
Thrombocytopenia gene panel
Thrombophilia gene panel

Immunology
IKBKG deletion/duplication analysis
Primary immunodeficiency gene panel
Severe combined immunodeficiency gene panel

Metabolic Disorders

Fatty acid oxidation disorders gene panel
Glycine encephalopathy gene panel
Glycogen storage disorder gene panel
Glycosylation (CDG) disorders gene panel
Methylmalonic aciduria gene panel
Organic acidemia gene panel
Leigh syndrome & mitochondrial encephalopathy gene panel
Ornithine transcarbamylase deficiency (OTC) deletion/duplication analysis
Lysosomal storage disorder gene panel
Urea cycle defects gene panel

Nephrology

Alport syndrome gene panel
Bartter syndrome gene panel
Meckel Gruber syndrome gene panel
Nephrotic syndrome gene panel
Polycystic kidney disease gene panel
Primary hyperoxaluria gene panel
Ciliopathy gene panel

Neurology - Neuromuscular

Arthrogryposis & congenital myasthenic syndrome gene panel
Charcot-Marie-Tooth and sensory neuropathies gene panel
Muscular dystrophy & congenital myopathy gene panel
Duchenne muscular dystrophy (DMD) gene analysis
Duchenne Muscular Dystrophy (DMD) deletion/duplication
Myotonia congenita gene panel
PMP22 deletion/duplication analysis
Spinal Muscular Atrophy (SMN1) gene analysis
Spinal Muscular Atrophy (SMN1/SMN2) deletion/duplication

Neurology-Epilepsy

Aicardi-Goutieres syndrome gene panel
Neuronal migration disorder gene panel
comprehensive epilepsy gene panel
Rett syndrome gene panel

Neurology - Movement Disorders

Ataxia-telangiectasia (ATM) gene analysis
Ataxia-telangiectasia (ATM) deletion/duplication
Dystonia gene panel
Early-onset juvenile parkinsonism gene panel
Hereditary spastic paraplegia gene panel
Hyperekplexia gene panel
Neurofibromatosis (NF1 and NF2) gene analysis
Neurofibromatosis type 1 (NF1) deletion/duplication
Neurofibromatosis type 2 (NF2) deletion/duplication

Neurology – Neurodegenerative

Adrenoleukodystrophy (ABCD1) gene analysis
Joubert syndrome gene panel
Leukodystrophy gene panel
Metachromatic leukodystrophy gene panel
Neurodegeneration with brain iron accumulation (NBIA) gene panel
Neurodegeneration with brain iron accumulation 2B (PLA2G6) deletion/duplication analysis
Pantothenate kinase-associated degeneration (PANK2) deletion/duplication analysis

Oncology

Hereditary cancer panel
Breast Cancer panel

Ophthalmology

Leber congenital amaurosis gene panel
Optic atrophy gene panel
Retinal degeneration gene panel
Congenital cataract gene panel
Cone-rod dystrophy gene panel
Retinitis Pigmentosa gene panel

Rare disorders

Bardet-Biedl syndrome gene panel
Cornelia de Lange syndrome gene panel
DiGeorge syndrome deletion/duplication analysis
Cystic Fibrosis (CFTR) gene analysis
Cystic fibrosis (CFTR) gene deletion/duplication
Noonan syndrome gene panel
Prader-Willi/Angelman syndrome deletion/duplication
Stickler syndrome panel

Skeletal disorders

Skeletal dysplasia gene panel
Osteogenesis imperfecta gene panel
Osteopetrosis gene panel
Achondroplasia (FGFR3) gene analysis

New born ICU genetic screening panel

New born genetic testing screening panel
New born genetic testing diagnostic panel

Reproductive panel

Infertility gene panel
Arrested embryo development gene panel
Recurrent pregnancy loss gene panel

Pulmonology

Pulmonary Artery Hypertension (PAH) gene panel
Cystic Fibrosis (CFTR) gene analysis
Cystic fibrosis (CFTR) gene deletion/duplication
Primary ciliary diskinesia gene panel
Surfactant metabolism dysfunction gene panel

REPORT EXAMPLES

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WHOLE EXOME SEQUENCING DIAGNOSTIC REPORT

Patient Information		Sample Information		Clinic Information	
Patient Name:		Specimen Type:	EDTA-Blood	Unique Patient ID:	200314
Patient DOB:	November 14, 2019	Date of Collection:	NA	Referral Clinic:	
Gender:	Male	Receipt Date:	March 10, 2020	Clinic Location:	Jordan
Lab Code:	WED-20M0009	Report Date:	May 4, 2020	Referral Physician:	

CLINICAL INDICATION:

Mohammad Ibrahim Al Maraba'ah is born of a consanguineous marriage. He presented with clinical symptoms of hypertrophic cardiomyopathy with severe left ventricular impairment, protruded tongue, axial and peripheral hypotonia and liver problem. He is suspected to be affected with Pompe disease. There is a positive family history of sibling death due to cardiomyopathy.

GENETIC ANALYSIS SUMMARY:

A homozygous pathogenic variant in the *GAA* gene was identified in the index. The *GAA* gene is associated with autosomal recessive glycogen storage disease II. The result is consistent with a possible genetic diagnosis of Pompe disease.

VARIANT(S) RELATED TO THE PHENOTYPE

GENE	GENOMIC POSITION (GRCh37)	NUCLEOTIDE CHANGE (TRANSCRIPT)	PROTEIN CHANGE (EFFECT)	ZYGOSITY	SNP IDENTIFIER	MINOR ALLELE FREQUENCY*	DISORDER (OMIM, INHERITENCE)	VARIANT CLASSIFICATION **
GAA (*606800)	chr17: 78081379delT	c.716delT (NM_000152.4)	p.Leu239ArgfsTer29 (Frameshift)	Homozygous	rs1555599594	N/R	Glycogen storage disease II (#232300, AR)	Pathogenic

*Highest frequency present in Exome Aggregation Consortium database (ExAC),1000Genome project (1000G), or gnomAD **based on ACMG Guidelines; AD: Autosomal Dominant, AR: Autosomal Recessive, XLD: X-Linked Dominant, XLR: X-Linked Recessive; N/R: Not Reported; N/A: Not available

VARIANT INTETPRETATION:

By whole exome sequencing, a homozygous frameshift variant in the *GAA* gene (c.716delT, p.Leu239ArgfsTer29) was identified. This variant is present in exon 4/20 and the reference region is conserved across the species. This variant has previously been reported in a patient affected with glycogen storage disease type II (PMIDs: 14695532). This frameshift variant is not present in population databases (1000 genomes and gnomAD). According to the recommendations of the ACMG, this variant is classified as 'Pathogenic'.

Glycogen storage disease II, an autosomal recessive disorder, is the prototypic lysosomal storage disease. In the classic infantile form (Pompe disease), cardiomyopathy and muscular hypotonia are the cardinal features; in the juvenile and adult forms, involvement of skeletal muscles dominates the clinical picture (PMID: 6360103).

In classic cases of Pompe disease, affected children are prostrate and markedly hypotonic with large hearts. The tongue may be enlarged. Although the enzyme is deficient in all tissues, muscle weakness and heart involvement are the most common features. The liver is rarely enlarged, except as a result of heart failure, and hypoglycemia and acidosis do not occur as they do in glycogen storage disease I (#232200). Death usually occurs in the first year of life in the classic form of the disorder and cardiac involvement is striking. Indeed, Pompe (1932) reported this condition as 'idiopathic hypertrophy of the heart,' and 'cardiomegalia glycogenica' is a synonym.

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CHROMOSOMAL MICROARRAY ANALYSIS TEST REPORT

Patient Information		Sample Information		Clinic Information	
Patient Name:	XXXX	Specimen Type:	EDTA-Blood	Unique Patient ID:	XXXX
Patient DOB:	XXX	Date of Collection:	NA	Referral Clinic:	XXX
Gender:	Female	Receipt Date:		Clinic Location:	UAE
Lab Code:	I3E-20A0008	Report Date:		Referral Physician:	XXXX

CLINICAL INDICATION / HISTORY:

Ms. XXXX is consanguineously married. She has a history of 3 early miscarriages (two miscarriages from spontaneous pregnancy, another early miscarriage from IUI treatment) and 4 IVF in other clinic, always with bad embryo development or arrested embryos. She has been evaluated for pathogenic copy number variations (CNV) by chromosomal microarray analysis.

ARRAY TYPE:

Affymetrix 750k Ingles.

RESULTS AND ANALYTICAL INTERPRETATION:

Array-CGH formula according to ISCN 2016: arr(1-22,X)x2. The referred sample shows a FEMALE SEX genomic pattern.

We have not detected DNA copy number changes in the DNA sample.

We have additionally detected several large regions of homozygosity (totally 74.46 Mb), encompassing 3% of the whole genome. According to the references[1], an individual with ~3% homozygosity could indicate that this individual is an offspring of two consanguineal parents which could mean a fourth degree of consanguinity between them. This result raises the possibility of a recessive disorder with a causative and mutated gene located within one of these regions (listed below).

Chromosome	Cytoband Start	Cytoband End	Size (Mb)
1	q41	q42.13	8.51
1	q43	q44	5.65
2	q33.3	q35	10.48
4	q12	q13.1	8.23
5	q22.3	q31.1	17.74
8	q23.2	q24.11	7.16
11	p15.5	p15.4	5.16
X	p22.13	p21.3	6.35
X	p11.23	p11.22	5.18

A detailed analysis of the 11p15 region concluded that there were some genes and regions involved (*CDKN1C* (*600856) and *KCNQ1* (*607542), including ICR2 region) whose abnormal imprinting could be cause of Beckwith- Weidemann syndrome. If this syndrome was suspected phenotypically in the probandus, a MS-MLPA/methylation analysis should be recommended to confirm the methylation of ICR2 associated to Beckwith-Wiedemann.

SUMMARY AND COMMENTS:

We have not detected any CNV that could explain the phenotype of the index. Several large regions of homozygosity were detected, indicating that the proband is an offspring of consanguineal parents at third degree. One of these homozygous regions was located at 11p15, including the Beckwith-Wiedemann methylation targets.

REPORT EXAMPLES

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Gender:	Male	Receipt Date:	March 10, 2020	Clinic Location:	Jordan
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CLINICAL INDICATION:

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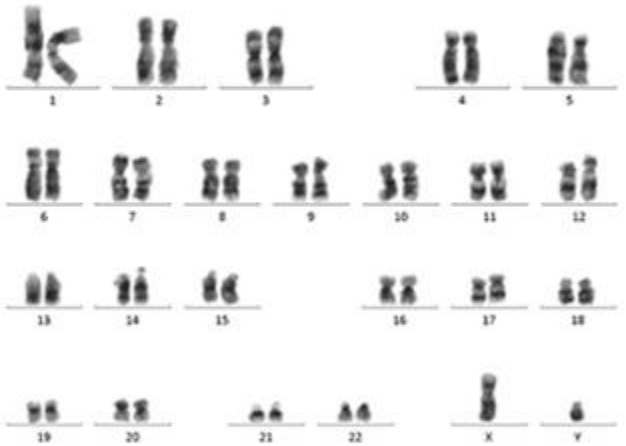
Unit 501 and 502, Building 40,
Dubai Health Care City,
P.O. Box 66066 Dubai UAE

Cytogenetic Analysis Report

PATIENT NAME: XXXXXX
Date of Birth: XX/XX/XXXX
MRN No: 7621 M
Specimen: peripheral blood lymphocytes
Report Date: 30/03/2020
Reception Date: 11/02/2020
Methodology: peripheral blood lymphocyte cell culture and GTG banding.



Karyotype: 46,XY

Results: Chromosomal analysis (GTG-Banding with 500 band resolution) revealed an apparently normal male karyotype in all the metaphases analysed.

Note: The cytogenetic result in samples of peripheral blood does not exclude the presence of non-detectable abnormalities due to limitations inherent of the technique itself, such as; low frequency mosaicism and cryptic structural abnormalities (microdeletions, microduplications and telomeric translocations), demonstrated by other molecular techniques. In this study normal variants without clinical implication are not specified.

SIGNATURE
Laboratory Director
(Validated at Igenomix)

COMPANY PROFILE

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WORLDWIDE REFERENCE COMPANY IN GENETICS SERVICES
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23 GENETIC LABORATORIES WITH PRESENCE IN MORE THAN 80 COUNTRIES

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SPAIN HEAD QUARTER

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A GROWING FAMILY OF 500 EMPLOYESS 20% PhDs

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AN INTERNATIONAL TEAM EXPERIENCED AND REPUTED (450 EMPLOYEES AND 20% PhD)

Industry Experience

22 
Carmen Rubio, PhD
Lab Director PGT-A H.Q.

11 
Ana Cerveró, PhD
Lab Director PGT-A H.Q.

15 
Julio Martin, PhD
Director of Applied
Clinical Development

17 
David Blesa
Product Development
Directo

Industry Experience

22 
Javier Garcia Planells
Clinical Director and Scientific
Rare Diseases

12 
Marcia Riboldi, PhD
Lab Manager Brazil

04 
Vandana Sharma, PhD
Lab Manager India

14 
Rupali Chopra, PhD
Lab Dir. Middle East

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