Sous L'égide de la Faculté de Médecine de Sfax (FMS)

L'Association du Développement de la Recherche, de l'Innovation et de l'Insertion des Compétences (ADRÏC)

Le Laboratoire de Génétique Moléculaire Humaine de la Faculté de Médecine de Sfax (LGMH)

Le Service de Génétique Médicale de l'Hôpital Hédi Chaker de Sfax (SGM)

Le Laboratoire de recherche Toxicologie Microbiologie Environnementale et Santé (TMES)

Organisent :

En collaboration avec

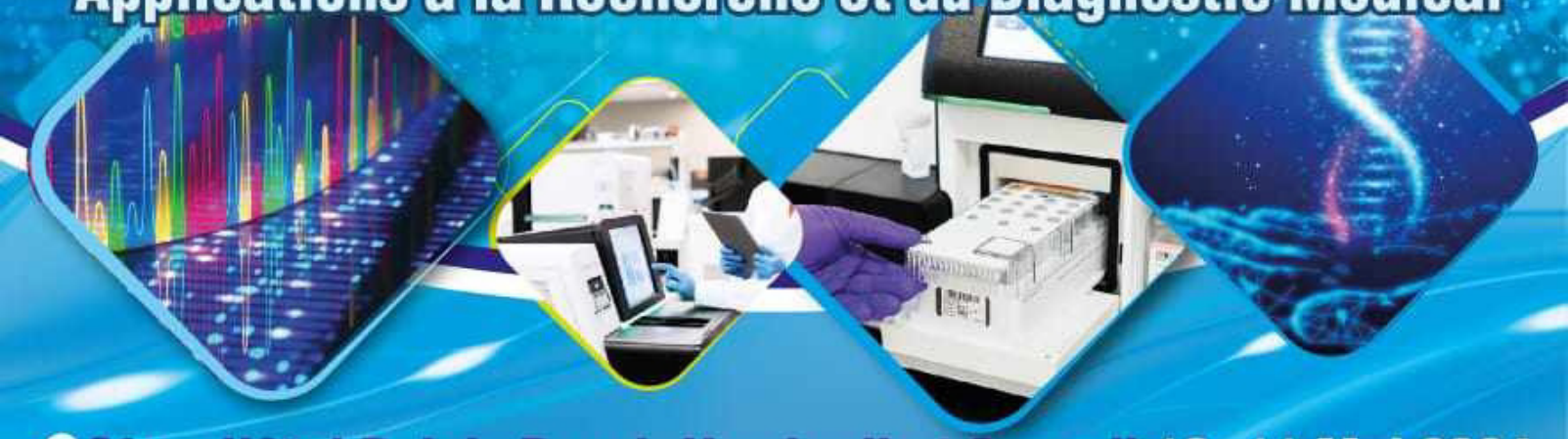
Le Centre de Biotechnologie de Sfax (CBS)

Un Colloque Scientifique

Intitulé

NOUVELLE GENERATION DE SEQUENÇAGE (NGS) :

Applications à la Recherche et au Diagnostic Médical



Sfax, Hôtel Palais Royal, Vendredi et Samedi 10 -11 Mai 2024

Ce colloque réunira : Enseignants, Chercheurs, Médecins, Doctorants, Ingénieurs et Techniciens

◆ Frais d'inscription

Les frais de participation au colloque avec ou sans poster , incluent l'inscription, documents, pause café et Brunch : 190 DT Hors Taxe (TVA 19%)

◆ Contact

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Next Generation Sequencing (NGS): Research and Medical Diagnosis Applications

May 10-11, 2024; Palais Royal Hotel - Sfax - TUNISIA

PROGRAM

1st Day : Friday May 10, 2024

13:30-14:30	**Welcoming Guests, Networking Registration,**
	POSTER DISPLAY AND VIEWING
14:30-15:00	**Inaugural Conference** : The Next Generation Sequencing Technologies : A Short Overview. ***Assoc. Pr Hassen HADJ KACEM*** *(University of Sharjah, UAE)*
	Session 1 : NGS APPLICATIONS IN RESEARCH
	Moderators : Pr Hammadi AYADI and Pr Raja GARGOURI
15:00-17:30	**CONFERENCES AND DISCUSSION**
15:00-15:30	Qatar Genome Odyssey: Navigating from Research to Precision Healthcare Realization. ***Dr Hamdi MBAREK*** *(QATAR Genome Project, Qatar)*
15:30-16:00	Tunisian Human Genome Project. ***Dr Yosr HAMDI*** *(Pasteur Institute, Tunis, Tunisia)*
16:00-16:30	**COFFEE BREAK AND POSTER SESSION**
16:30-17:00	Metagenomic Analysis of the Human Microbiota in Tunisian Individuals. ***Pr Leila KESKES*** *(MICAfrica Project, Faculty of Medicine of Sfax, Tunisia)*
17:00-17:30	From Next Generation Sequencing Microorganisms Towards Eco Friendly Biotech Based Products. ***Pr Slim TOUNSI*** *(NGS-4ECOPROD Project, Center of Biotechnology of Sfax, Tunisia)*
17:30-18:00	**ORAL COMMUNICATIONS AND DISCUSSION**
17:30-17:45	**Communication 1** : Genomic characterization of clinical Salmonella Typhimurium DT104 isolates. ***Sonia KTARI*** *(Faculty of Medicine of Sfax, Tunisia)*
17:45-18: 00	**Communication 2** : Exploring the Taxonomic Diversity of Bacterial Communities in Sidi Ayed Hot Spring, West Algeria, Through Metagenomic Investigation and Non-Culturomics Methods. ***Marwa AIRECHE*** *(Higher School of Biological Sciences of Oran, Algeria)*
18:00-18:30	**GENERAL DISCUSSION AND FIRST DAY CLOSING**

2nd Day : Saturday May 11, 2024

	Session 2 : NGS APPLICATIONS IN DIAGNOSIS : PUBLIC SECTOR
	Moderators : Pr Mouna TURKI and Pr Hassen KAMOUN
09:00-11:30	**CONFERENCES AND DISCUSSION**
09:00-09:30	Evolution of Medical Genetics at Charles Nicolle Hospital : Challenges, Implementation, and the Impact of Next-Generation Sequencing on Patient Care. ***Pr Ridha M'RAD*** *(Charles NICOLLE Hospital, Tunis, Tunisia)*
09:30-10:00	From Tackling Gaps in the Implementation of NGS to the First on-site Experiments. ***Pr Dorra H'MIDA*** *(Farhat HACHED Hospital, Sousse, Tunisia)*
10:00-10:30	Mutations Causing Mitochondrial Diseases in Tunisian Patients identified by NGS : Molecular and Structural Investigations. ***Pr Faiza FAKHFAKH*** *(Faculty of Sciences of Sfax, Tunisia)*
10:30-11:00	Advancing Health Through African Genomics: on The Way to the 3 Millions Genomes Dream. ***Pr Ahmed REBAI*** *(Center of Biotechnology of Sfax, TUNISIA)*

11:00-11:30	Feedback and Challenges of Next Generation Sequencing in Neuro-Developmental Disorders. ***Assoc. Pr Ikhlas BEN AYED*** *(Hedi CHAKER Hospital, Sfax, TUNISIA)*
11:30-12:30	**ORAL COMMUNICATIONS AND DISCUSSION**
11:30-11:45	**Communication 1** : **Development of a custom high-throughput sequencing panel for the diagnosis of Developmental and Epileptic Encephalopathies in the Tunisian population**. ***Mariem BEN SAID*** *(Center of Biotechnology of Sfax,Tunisia)*
11:45-12:00	**Communication 2** : **Etiological diagnostic contribution of Exome sequencing in neurodevelopmental disorders in a cohort of 100 Tunisian Patients**. ***Sana KAROUI*** *(Mongi SLIM Hospital, La Marsa, Tunisia)*
12:00-12:15	**Communication 3** : **Prognostic significance of *IKZF1* gene alterations in Tunisian patients with acute lymphoblastic leukemia**. ***Ameni BEDOUI*** *(Center of Biotechnology of Sfax, Tunisia)*
12:15-12:30	**Communication 4** : **Involvement of the intestinal microbiota in hypercholesterolemia and therapeutic challenge via probiotics**. ***Oumayma CHRIAA*** *(Faculty of Sciences of Sfax,Tunisia)*

Session 3 : ONCO-HEMATOLOGY SYMPOSIUM AND BRUNCH

Moderators : Pr Moez ELLOUMI and Pr Mouna MNIF

12:30-14:00	**BRUNCH, NETWORKING AND POSTER SESSION**
12:30-13:00	**Symposium** **NGS Onco-Hematology : Practical Interest** ***Assoc. Pr Imen FRIKHA*** and ***Pr Moez MDHAFFAR*** *(Hedi CHAKER Hospital, Sfax, Tunisia)*
13:00-14:00	**Brunch**

Session 4 : NGS APPLICATIONS IN DIAGNOSIS: PRIVATE SECTOR

New Sequencing Technologies in Genetic Diagnosis : Feedback from Collaboration in the Private Sector

Moderators : Pr Souad ROUIS and Pr Tahya BOUDAWARA

14:00-14:15	***Sponsor Communication : HTDS***
14:15-14:30	***Sponsor Communication : ADVANCED***
14:30-16:00	**CONFERENCES AND DISCUSSION**
14:30-15:00	Sequencing Technologies and Data Analysis : ***Dr Ahmed Sahbi CHAKROUN*** *(RAN BioLinks Company, Tunis, Tunisia)*
15:00-15:30	Application of WES in Rare Genetic Diseases Diagnosis : ***Dr Mohamed Ali KSENTINI*** *(Private laboratory of Cytogenetics, Sfax, Tunisia)*
15:30-16:00	NGS and Oncology : ***Dr Mounira MEDDEB*** *(Private laboratory of Cytogenetics, Tunis, Tunisia)*
16:00-17:00	**AWARDS AND CLOSING**


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10 -11
Mai 2024

INAUGURAL CONFERENCE :

THE NEXT GENERATION SEQUENCING TECHNOLOGIES:

A SHORT OVERVIEW

Pr. Hassen HADJ KACEM

Speaker

- **2015-2024** : **Associate Professor in the Department of Applied Biology, College of Sciences, University of Sharjah**
- **2006-2012 : Associate Professor in the Centre of Biotechnology of Sfax (Tunisia).**
- **2005 : Young Researcher (PostDoc)in Cochin Institute, University René Descart, Paris V (France),**
- **2004 : Young Researcher in the Laboratory of Human Molecular Genetics; Faculty of Medicine of Sfax, Tunisia.**
- **2000 : Teaching assistant in the Faculty of Sciences of Sfax, University of Sfax (Tunisia).**

Department of Applied Biology, College of Sciences. University of Sharjah

hkacem@sharjah.ca.ae

Scopus 48 publications, h-index: 14

CONFERENCE TITLE :

QATAR GENOME ODYSSEY: NAVIGATION FROM RESEARCH TO PRECISION HEALTHCARE REALIZATION

Dr. Hamdi MBAREK

Speaker

Since 2019 : Research Partnerships Manager at Qatar Genome Program

Since 2017 : Asssistant Professor in Vrije Universiteit Amsterdam, Netherlands

2012-2017: Postdoctoral Fellow in the Faculty of Behavioural and Movement Sciences, Amsterdam, Netherlands

2015 : Teaching Human Molecular Genetics course in Vrije Universiteit Amsterdam

2010-2011: Postdoctoral Fellow, Institute of Medical Science, Human Genome Center, in The University of Tokyo; Japon

2006-2009: PhD thesis , GenHotel-EA3886, Evry-Val-d'Essonne University, Evry-Genopole, France

Research Partnerships Manager at Qatar Genome Program

hmbarek@qf.org.qa

Scopus **117 publications, h-index :41**

CONFERENCE TITLE :

TUNISIAN HUMAN GENOME PROJECT

Dr. Yosr HAMDI

Speaker

- **Since 2018 : Leader of Collaborative Alliance and the PerMediNA Consortium**
- **2015-2018: Cancer genetics in North African project**
- **2009-2014: Association of common genetic polymorphisms and breast cancer risk project**
- **2007-2009: Member of Breast Cancer and Oxidative Stress project**
- **2004: Member of the Human Genome project**
- **Expert and technical advisory Group on Genomics in the World Health Organization**

Pasteur Institute of Tunis, Biomedical Genomics and Oncogenetics Laboratory

yosr.hamdi@pasteur.utm.tn

Scopus **45 publications ; h-index: 13**

The Genome Tunisia Project: Towards a Data-Driven Healthcare System

Yosr Hamdi, PhD

Laboratory of Biomedical Genomics and Oncogenetics
Institut Pasteur de Tunis, Tunisia

Several discoveries in the field of human genetics have led to the foundation of modern molecular and personalized medicine.Here, we are presenting the Genome Tunisia Project that aims to determine the reference sequence of the Tunisian Genome in order to, ultimately, implement precision medicine in Tunisia.

The main goal of this initiative is to develop a healthcare systemcapable of incorporating Omics data for use in routine medical practice, enabling medical doctors to better treat, prevent and diagnose patients. Therefore,a multidisciplinary partnership involving Tunisian experts from different regions has come to discerning all requirements that would be of high priority to fulfill the project goals. One of the most urgent priorities is to determine the reference sequence of the Tunisian Genome.

To achieve these goals, hundreds of Tunisian Genomes have been already sequenced and data analysis is in progress. In addition, extensive situation analysis of the education programs, community awareness, training, appropriate infrastructure as well as the ethical and regulatory framework have been undertaken towards building sufficient capacity to integrate genomic medicine in the Tunisian healthcare system.

The Genome Tunisia initiative seeks to demonstrate the major impact that can be achieved by implementing National Human Genome Projects in LMICs to improvedisease management and treatment outcome. Additional effortsare now

made for the advancement of patient care by accelerating research and innovation, and supporting necessary changes in policy and regulation.

CONFERENCE TITLE :

METAGENOMIC ANALYSIS OF THE HUMAN MICROBIOTA IN TUNISIAN INDIVIDUALS

Pr Leila AMMAR KESKES

Speaker

- **2021-2024: Coordinator of MICAfrica project funded by the EC within H2020 program**
- **2021-2022: Tunisian National Contact Point in Horizon Europe**
- **2018-2020 : Tunisian National Contact Point in "Health" of H2020 and Horizon Europe (2021-2022)**
- **Since2013 : Senior Researcher and Director of LGMH at the Faculty of Medicine of Sfax- Tunisia**
- **Since 2009 : MD and Professor in Histology-Embryology at the Faculty of Medicine of Sfax- Tunisia**

Faculty of Medicine of Sfax, Human Molecular Genetics Laboratory (LGMH)

Leila.ammar@fms.usf.tn

Scopus 179 publications ; h-index 25

Metagenomic Analysis of the Human Microbiota in Tunisian Individuals (MICAfrica project)

Pr. Leila Ammar-Keskes

MICAfrica Collaborators: Pr. Radhouan Gdoura, Pr. Raja Gargouri, Pr. Hayet Sellami, Pr. Basma Mnif, Pr. Ali Amouri

MICAfrica is a Twinning project funded by the European Commission in the framework of H2020 Programme. It is a Coordination and Support Action (CSA) with the main objective to support the University of Sfax (USFAX) in Tunisia in developing skills in human microbiome analysis and becoming a leading force in the region. The collaboration with three European partners: the "Institut National de la Recherche Agronomique, Aix-Marseille University in France and the University of Florence in Italy helped the USFAX to increase its staff's technological capabilities, adopt standardised methods for collecting samples and analysing data and establish a consortium based on existing collaborations in Morocco, Algeria , Libya and Egypt.

During MICAfrica, the research activities focused on the use of NGS technologies to analyse the Human Microbiota and to study the relationship between the disruption of gut Microbiota called dysbiosis and Colorectal Cancer (CRC).

A pilote study involving 5 CRC patients and 5 controls and using total metagenomic analysis by Illumina and Oxford Nanopore NGS technologies showed the implication of few bacterial genera and species in CRC.

Also a comparative study aiming to compare the efficiency of four DNA extraction methods from human stool samples for high-throughput sequencing by Nanopore technology, revealed considerable variations among DNA extraction methods and showed a highest DNA recovery with the extraction method based on enzymatic digestion which emerged as the most effective extraction method.

Finally, a metagenomic analysis performed on 68 formalin-embedded paraffin fixed tissues from tumoral (n = 33) and healthy mucosa (n = 35) collected from 35 CRC Tunisian patients revealed the presence of archaeal sequences and highlighted an enrichment in *Halobacteria* in the tumor tissues with a significant association between *Halobacteria* and tumor tissues in

patients with CRC. The findings demonstrated in addition the feasibility of archaeome analysis from formol fixed paraffin-embedded (FFPE) tissues.

CONFERENCE TITLE :

FROM NEXT GENERATION SEQUENCING MICROORGANISMS TOWARDS ECO FRIENDLY BIOTECH BASED PRODUCTS

Pr. Slim TOUNSI

Speaker

- **PI/CoPI of several Joint Research Projects (Bilateral and Multilateral: NGS-4ECOPROD, H2020, ARIMNet, ERANETMED, COST,..).**
- **2018: The presidential award for the best Lab in Tunisia.**
- **2015: the second award of Univenture/Wiki Start-Up**
- **2014: The regional award of innovative project/PASRI-ANPR**
- **2006: Second PhD, Biology sciences and Biotechnology at the Higher Institute of Biotechnology of Monastir-Tunisia**
- **2002: Biology Engineering Doctor's degree at the School of Engineer of Sfax-Tunisia.**

General Director of the Biotechnology Center of Sfax

slim.tounsi@cbs.rnrt.tn

Scopus 132 publications ; h-index: 30

Abstract : Slim Tounsi

Biotechnologydevelops and appliesmicroorganisms for the production of bioproductswithindustrialinterest. Nextgeneration-sequencing (NGS) plays an increasingly important role in improving and acceleratingmicrobialstraindevelopment for existing and novelbioproducts via screening, gene and pathwaydiscovery, metabolic engineering and additionaloptimization and understanding of large-scalemanufacturing. NGS technologies have become main stream due to an important reduction in costs and an increase in sequencequality. However, data analysisstillrequiressubstantialbioinformatics expertise and adaptation to specificpurposes due to the large datasets, data management and infrastructure supporting an NGS researchfacility. These challenges are particularly relevant in the research centers of the widening countries such as the case of the Centre of Biotechnology of Sfax (CBS) in Tunisia. In line with EU orientations and twinningrequirements, NGS-4-ECOPROD aimsto: (i) improvecreativity, excellence capacity, and resources, (ii) raise the reputation, research profile, and attractiveness, and (iii) strengthresearch management capacities and administrative skills in the CBS, throughaddressing the existing gaps in the field of NGS. NGS-4-ECOPROD projectwillallow CBS to exploit its NGS platform to develop original biotechnologyproducts (extremozymes, sporeless biopesticides, antioxidant-ergothioneine, and bioplastics) useful as eco-friendly alternative to chemicalsones. This willbeachievedthanks to the tight collaboration and networking activities (staff exchanges, workshops, conferences, training youngresearchers, and summerschoolactivities) between the CBS and twoleadingwellrenownedresearch EU Partners in the field of NGS namely the University Claude Bernard Lyon 1 in France and the Georg August University of Göttingen in Germany. The NGS-4-ECOPROD willultimatelyadopt an integrated communication and disseminationstrategywithopenness on the socio-economicsectors.

CONFERENCE TITLE :

EVOLUTION OF MEDICAL GENETICS AT CHARLES NICOLLE HOSPITAL: CHALLENGES, IMPLEMENTATION, AND THE IMPACT OF NEXT-GENERATION SEQUENCING ON PATIENT CARE.

Pr Ridha M'RAD

Speaker

- **Medical Genetics professor in the Medicine Faculty of Tunis,**
- **Coordinator: CEC for Medical Genetics and Genomics**
- **Member of the Human Genetics research Laboratory at Faculty of Medicine of Tunis**
- **Head of Service: Congenital and Hereditary Diseases Department at Charles Nicolle Hospital**
- **President of the Tunisian Society of Medical Genetics STGM**

Faculty of Medicine of Tunis

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Scopus 97 documents; h-index 16

"Évolution de la génétique médicale à l'Hôpital Charles Nicolle : Défis, réalisation et impact du séquençage nouvelle génération sur la prise en charge des patients" "Evolution of Medical Genetics at Charles Nicolle Hospital: Challenges, Implementation, and the Impact of Next-Generation Sequencing on Patient Care"

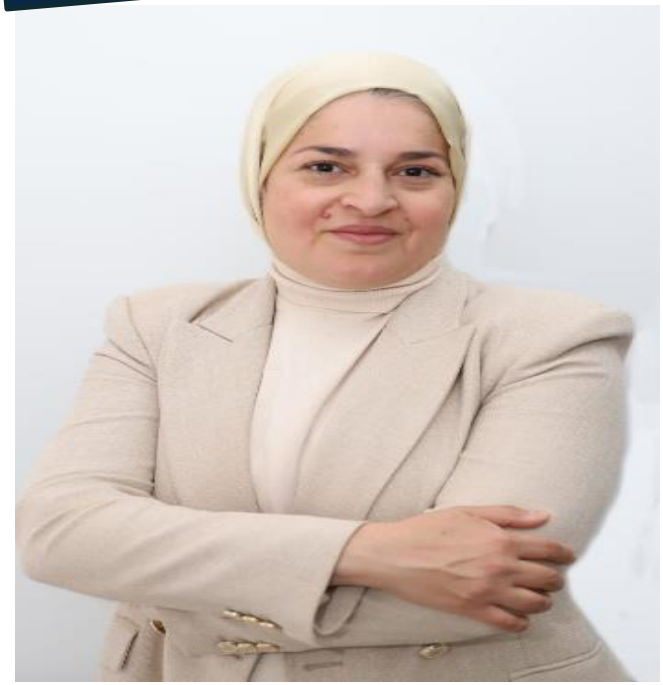
Summary:

Rapid technological advances in medical genetics, particularly in the field of DNA sequencing, have had a significant impact on healthcare. In recent decades, technological progress has enabled detailed mapping of the human genome, leading to new diagnostic approaches for genetic diseases and cancers. These developments have transformed medical practice by allowing healthcare professionals to offer personalized treatments and more accurate predictions of genetic risks. However, this rapid progression presents significant challenges. The organization of medical genetics within hospitals must adapt to new technologies, requiring substantial investments in sequencing platforms and specialized training for professionals. Ensuring the quality of analyses and interpretations is essential to avoid diagnostic errors. Furthermore, the line between scientific progress and commercial innovation is fine, necessitating a rigorous ethical approach to avoid misuse. Another critical aspect is balancing the installation of next-generation sequencing (NGS) platforms in hospitals with public health policy objectives, given the still-high cost of genetic testing. These high costs raise concerns about healthcare affordability and equity, as not all hospitals and patients have the same resources to access these technological advancements. Additionally, treatments indicated by genetic testing can be expensive, increasing the financial burden on patients and healthcare systems. Thus, a sustainable economic model is required to enable the installation of NGS platforms without compromising the quality of care. These challenges underscore the importance of ensuring that access to cutting-edge medical genetics technology is not limited to a privileged few but is part of an inclusive public health policy. This requires strategic partnerships among hospitals, biotechnology companies, and health authorities to reduce costs while maintaining high standards. Reconciling technological advancements and their high costs with a broader vision of public health is crucial. Coordinated efforts are needed to ensure that innovations in medical genetics contribute to a fair healthcare system, where technological progress benefits everyone without compromising the quality of patient care.

CONFERENCE TITLE :

FROM TACKLING GAPS IN THE IMPLEMENTATION OF NGS TO THE FIRST ON-SITE EXPERIMENTS.

Pr. Dorra H'MIDA



Speaker

- **February 2024 :Founder President of the Tunisian Society of Human Genomics TSHG**
- **Since 2012: Membre of Orphanet Genetic Advisory Board. France, Paris.**
- **2015-2019 : Associate Professor in the Laboratory of Cytogenetics and Molecular Genetics and Human Reproductive Biology Farhad HACHED Sousse .**
- **2009-2015: Hospital University Assistant.**
- **2003-2009 :Medical resident .**

Genetics Department, Farhat HACHED hospital,

dorrahmida@yahoo.fr

Scopus 36 documents; h-index 15

Organisateur : ADRIC
Manifestation scientifique : SEQUENÇAGE NOUVELLE GENERATION (NGS) : Applications à la Recherche et au diagnostic médical. 10 et 11 Mai Sfax

Conference:

Professor Dorra H'MIDA, MD, PhD, Department of Genetics, Farhat HACHED university Hospital of Sousse, Faculty of Medicine, University of Sousse, Tunisia.

Title: Farhat HACHED University Hospital Experience: from tackling gaps in the implementation of NGS to the first on-site experiments

Abstract:
Background: Since its establishment during the 1990s, the department of Genetics at Farhat HACHED University hospital aimed at providing optimal genetic testing and counselling for referred patients and their families. Over the years, technical capabilities have evolved, starting from cytogenetics and low-throughput molecular genetic techniques, leading to High throughput tests (HT) and NGS adoption.

Content:
The NGS testing adoption started during the 2010s, through testing carried out abroad, especially for cancer hereditary predisposition carrier patients. This activity remained at a minimum level as these tests are not refunded by the health insurance system and the NGS technology was not yet implemented in Tunisian public health institutions. Indeed, only a few patients who can afford the high costs of these tests had access to them, despite their significant value in clinical decision-making.
Despite these financial limitations, Tunisian patients demand for HT testing timidly increased over the years for 2 of the most mature areas for clinical application of genomics: oncology and rare genetic conditions. The large majority of conducted tests consisted of gene panels, with few WES. In 2016, thanks to a fruitful collaboration in frame of a VRR program led by the Centre of Biotechnologies of Sfax (CBS), we provided to our patients, the first Tunisian made NGS panels.
In 2021, through a grant, we managed to conduct 300 WES abroad. Patients were selected for 3 clinical presentations: hereditary cancer predisposition, syndromic intellectual deficiency, and genodermatosis.
In 2023, was implemented the NGS technology in our department. Although no specific budget was yet allocated for the routine NGS diagnostic, our team took advantage of a national research program (PRF "DINS Socio-Genomics") to raise funds for clinical exome analysis for non-syndromic intellectual deficiency. To date,

Organisateur : ADRIC
Manifestation scientifique : SEQUENÇAGE NOUVELLE GENERATION (NGS) : Applications à la Recherche et au diagnostic médical. 10 et 11 Mai Sfax
our team is in the development phase of on-site NGS analysis in frame of this last project.

Summary: Despite genomics' success in transforming patients' care, few diagnostic funds are allocated for this aim and research funds are often utilized for diagnostic purpose. Integration of genomics into routine healthcare requires awareness among physicians and insurance providers. Given the financial capacity of Tunisian citizens, few people are able to afford the relatively high costs of genetic analyses. Consequently, it is essential at least for NGS panels and exome sequencing to be recognized by the Health Insurance system whenever there is a significant value in clinical decision-making.

CONFERENCE TITLE :

MUTATIONS CAUSING MITOCHONDRIAL DISEASES IN TUNISIAN PATIENT IDENTIFIED BY NGS: MOLECULAR AND STRUCTURAL INVESTIGATIONS

Pr. Faiza FAKHFAKH

Speaker

- **Head of Molecular and Functional Genetics laboratory, Faculty of Sciences of Sfax**
- **Mai 2022: Member of the Task force on Genetic Literacy of International league against Epilepsy (ILAE).**
- **2017-2021: Member of the Task force on advocacy for research of International league against Epilepsy (ILAE).**
- **Since 2016: Member of the executive board of the Tunisian Society of Biological Sciences**
- **2003-2012 :Head of Human Molecular Genetics Laboratory at the Faculty of Medicine of Sfax**

Faculty of Sciences; University of Sfax

faiza.fakhfakh02@gmail.com

Scopus 168 documents, h-index 22

Mutations causing mitochondrial diseases in Tunisian patients identified by NGS : Molecular and structural investigations.

Pr Faiza Fakhfakh : Laboratory of Molecular and Functional Genetics (LGMF), Faculty of Science of Sfax, University of Sfax

Mitochondrial dideases include multiple varieties of pathologies having as common denominator a dysfunction of the mitochondrial respiratory chain resulting in energy deficiency. They are charachetrized by a high clinical and phenotypic expression heterogeneity which associates dysfunction of variety of tissues with high energy demand (muscle, brain , heart , kidney …) .The diversity of function of mitochondria and its tissular distribution as well as the double genetic origin of its structural and functional proteins reflect the phenotypic and genetic complexity of mitochondrial disease. In mitochondrial diseases all mode of genetic transmission are possibleincluding maternal (mitochondrial DNA /mtDNA) or mendelian transmissions (nuclear DNA /nDNA) with more than 1000 genes which are potentially involved. Thus in these diseases, mutation could effect either mitochondrial genes (mtDNA) or nuclear genes encoding proteins involved in mitochondrial functions.
We report the molecular investigation by next generation sequencing (NGS) of patients with clinical features suggestive of mitochondrial diseases. In addition bioinformatic analyses were carried out as well as mtDNA deletion screening and copy number quantification in the blood of pattients. NGS revealed several mutations in genes related to the synthesis and maintenance of mtDNA (POLG, *TYMP*, OPA1 genes), to the translation of mitochondrial proteins (*AARS*2, KARS, *EARS*2 and *DARS*2 genes) and in genes encoding proteins involved in the assembly of respiratory chain or in mitochondrial membrane transport (*NDUFS*4, *SLC19A*3 and *SERAC* genes). Result of mtDNA quantification revealed abnormal mtDNA copy number in some patients with depletion (decrease in the mtDNA copy number) particularly in patients haboring mutation in genes related to replication and maintenance of mtDNA. In addition structural 3D revealed effect of identified mutations which caused conformational changes in mutated structure compared to the wild type and could therefore alter flexibility and stability ofproteins .Molecular Docking performed for some mutations revealed their effect on imparing protein/protein or protein /ligand interactions leading to partial or complete loss of function .These investigations could be valuable to support experimental functional studies for a better understanding of physiopathological mechanisms leading to mitochondrial diseases.

CONFERENCE TITLE :

ADVANCING HEALTH THROUGH AFRICAN GENOMICS: ON THE WAY TO THE 3 MILLIONS GENOMES DREAM

Pr. Ahmed REBAI

Speaker

- **2009-2018: Director of the Laboratory of Molecular and Cellular Screening Processes at the Centre of Biotechnology of Sfax.**
- **2008/2010 and 2019: Invited professor in Biostatistics at LeMans University (France)**
- **Invited lecturer at several international workshops in Bioinformatics/Biostatistics**
- **Expert member/evaluator/consultant in different national and international bodies (Data and Biospecimen Access Committee of the H3Africa consortium (2016); the PRIMA foundation (Spain, 2019), the AREF foundation (UK; 2020); the Science for Africa Foundation (Kenya; 2022).**

Molecular and Cellular Screening Processes Laboratory, Centre of Biotechnology of Sfax

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Scopus 275 documents, h-index: 40

Sharing of Genomics Data: Experiences of a pioneering access committee in Africa

Ahmed Rebai

Laboratory of Molecular and Cellular Screening Processes, Centre of Biotechnology of Sfax, University of Sfax, Tunisia

Email address: ahmed.rebai@cbs.rnrt.tn

Abstract:

With Africa entering the genetic characterization space, and the resultant generation of whole exome or genome sequence data for thousands of individuals, facilitating responsible access to genomic data is an ethical and scientific imperative. Generating genomic data from African populations has a significant benefit to the broader scientific community due to extensive genetic diversity observed in African genomes.Whilst it is in the best interests of Africans and their health that genomic data be accessible to researchers and health care professionals across the global community, access needs to be both facilitated and controlled.

Access to genomics data can be regulated via three possible approaches: (a) Unrestricted or public access: data are completely open and accessible for consultation and public download. (b) Registered access: requires the registration of the user to the repository before being able to download the data. (c) Controlled access: is the most stringent data access approach and requires credentialed users to apply for access to data through an intermediary body which evaluates requests and allows access if a request complies with stated prerequisites.

In controlled access, data access requests (DARs) are evaluated by a Data Access Committee (DAC). A DAC is a body composed of individuals with diverse expertise, who is responsible for data release to external requestors based on guidelines and policies which have been defined by the parties that collected the samples, produced the data and funded the process, whilst respectingthe regulations of the country or continental region from which the samples originated. DACs can be either independent of the institution that produced the data, or institutional, each scenario presenting advantages and disadvantages. It has been recognized that DACs play important roles in both promotion of data sharing and protection of all stakeholders (data subjects, communities, data producers, institutions where researchers are based, the country or even continent of affiliation) by encouraging secondary uses of data.

In this conference, I willshare the experience of the Human Heredity and Health for Africa (H3Africa) DAC in assessing and evaluating DARsfor the genomics data that have been generated under the H3Africa project (https://h3africa.org/). I will also address the current challenges of setting up data sharing bodies (DACs) responsible for data access to whole genome sequences (for over 1000 individuals) generated under the pan-African Network AGenDA, involving 10 countries (including Tunisia and Libya. I willfinally discuss the ways in which the work of this (or similar) DBACs can be improved to achieve the goals of simultaneous stimulation of genomics data production in Africa, and the subsequent responsible wide sharing of these data.

The H3Africa DBAC has played a role in ensuring that African genomes are responsibly interrogated for the benefit of the human population. The experience of this DBAC provides a template upon which new projects, or consortia, can build. In addition, the experience of this DBAC is now available to other upcoming initiatives and can provide advice or support, so that there is coordinated access to African data that are beneficial to the broader scientific community. It is hoped that increased availabilityof African genomics data, and its access to global researchers and companies, could benefit not only Africans, but humanity at large.

CONFERENCE TITLE :

FEEDBACK AND CHALLENGES OF NEXT GENERATION SEQUENCING IN NEURO-DEVELOPMENTAL DISORDERS

Assoc.Pr. Ikhlas BEN AYED

Speaker

Associate Professor, Department of Medical Genetics; Hedi CHAKER Hospital, Sfax.
Member of the Laboratory of Molecular and Cellular Screening Processes, CBS, Sfax.
Coordinator of the KAFSS project
Member of Consortium on Developmental and Epileptic Encephalopathy (Egypt, Mauritania, Morocco, Saudi Arabia, Tchad, Tunisia)
Founding member of ATDE, TSHG, and STGM associations.
Member of ADREMED association

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Scopus 10 documents, h-index : 4


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SYMPOSIUM :

NGS ONCO-HEMATOLOGY: PRACTICAL INTEREST

Pr. Moez MDHAFFAR

Speaker

- **Since 2018: Professor in clinical hematology**
- **Since 2009 : Member of the LLC study group in Tunisia**
- **Since 2009 : Member of the Multiple Myeloma study group in Tunisia**
- **Since 2009: Member of the PTAI adult study group in Tunisia**
- **Since 2016 : Member of the Lymhomes study group in Tunisia**
- **2014-2017 : Member of the clinical hematology college and of the Medical Committee at Hedi CHAKER hospital, Sfax**

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Scopus 18 documents, h-index 6

SYMPOSIUM :

NGS ONCO-HEMATOLOGY: PRACTICAL INTEREST

Assoc.Pr. Imen FRIKHA

Speaker

- **Since 2021 : Associate professor in Hematology Department of Hedi CHAKER Hospital, Sfax, Tunisia**
- **Since 2020 : Member of the LLC study group in Tunisia**
- **Since 2019: Member of the study group for immunological thrombocytopenic purpura in adults in Tunisia**
- **Vice-president of the Tunisian hematology society term 2024 – 2027**

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Scopus 7 documents, h-index 3

NEXT GENERATION SEQUENCING IN MYELOID MALIGNANCIES

CLINICAL IMPLICATION

Pr Ag Imen FRIKHA, PrMoez MEDHAFFER, DrYossraFakhfakh, PrMoezElloumi

Department of Haematology of HediChakerHospital of Sfax, Tunisia

Abstract :

Hematologic malignancies represent a biologically and clinically diverse group of neoplasms arising from hematopoietic cells.These blood cancers have historically been more amenable to the adoption of new diagnostic technologies.The diagnosis has long included not only morphologic evaluation but also comprehensive immunophenotyping by flow cytometry and genetic characterization by conventional karyotype and fluorescence in situ hybridization. Next-generation sequencing (NGS) is not supplanting these tools but complementing them, adding additional precision to diagnostic hematopathology. In general, hematologic malignancies are divided based on the lineage of differentiation into myeloid and lymphoid neoplasms.In turn, the myeloid and lymphoid neoplasms can themselves be further subdivided into categories based on cell lineage, differentiation state, clinical features, and genetic findings.

Clinical NGS has been most widely adopted for myeloid malignancies, which will be the major focus of our presentation.Four major applications of somatic mutation data: (1) Facilitating diagnosis, (2) Informing prognosis, (3) Identifying targetable mutations, and (5) Monitoring disease.The subject of this presentation is to talk about clinical implication of NGS in myeloid malignancies.


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CONFERENCE TITLE :

SEQUENCING TECHNOLOGIES AND DATA ANALYSIS

Dr. Ahmed S CHAKROUN

- **2009-2024** : Co-Founder and COO RAN BioLinks Tunis, Tunisia
- **2009-2024** : Chief Executive Officer CarthaGenomics Advanced Technologies
- **2009-2004** : Laboratory associate in Pasteur Institute of Tunis
- **2004-2009**: Senior Engineer

Speaker

Private Biotechnology Laboratory, Tunis

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Abstract: Navigating the complexities of sequencing technologies and data analysis within a business context presents formidable challenges. In this lecture, we'll be delving into the pragmatic hurdles encountered by stakeholders in the private sector when implementing next-generation sequencing (NGS)-based genetic testing. Beyond technical considerations, our discussion encompasses a broader spectrum of concerns. Cost-effectiveness, quality assurance, system maintainability, and long-term sustainability emerge as pivotal focal points. Moreover, we explore the intricate interplay of the economic environment variables that must be carefully navigated. How can we strategically approach the value chain in this context? To what extent does fostering harmonized partnerships serve as a linchpin for sustained success and expansion? Join us as we unravel these critical questions and illuminate pathways towards operational effectiveness in NGS-based genetic testing within the private sector.


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CONFERENCE TITLE :

SEQUENCING TECHNOLOGIES AND DATA ANALYSIS

Dr. Mohamed Ali KSENTINI

Speaker

- **Since 2015: Director of a private laboratory specialized in genetics.**
- **Medical Doctor, Specialist in Medical Genetics; Faculty of Medicine of Sfax-Tunisia.**
- **Former Geneticist in the Department of Congenital and Hereditary Diseases, Charles Nicolle Hospital, Tunis and the Genetics Department, Hedi ChaKer Hospital, Sfax.**

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CONFERENCE TITLE :

SEQUENCING TECHNOLOGIES AND DATA ANALYSIS

Dr. Mounira MEDDEB

Speaker

- **Since 2002 : Director of a private laboratory specialized in genetics LABGEN**
- **Doctor in Medicine, Specialist in Medical Genetics , Faculty of Medicine of Tunis**
- **PhD in Human Genetics Université Paris VII**
- **Former Researcher Gustave Roussy Institute, France**
- **Former Head of Cytogenetics Department Private Laboratory, France**

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