“3RD CONFERENCE OF INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDIRC) “

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DIAGNOSIS AND TREATMENT ACCESS FOR PATIENTS WITH RARE DISEASES IN WEST AFRICA

Hawa Dramé - President of FITIMA
BIOGRAPHY

AFM (French Association against the Myopathies)

- ALLIANCE MALADIES RARES (Collective of French Associations) fighting against rare diseases: as assistant acting general
- EURORDIS (European Organization for Rare Diseases): as therapeutic development coordinator and expert at the COMP (orphan drugs committee of European Medicine Agency)
- ROAMY (West African Organization against the Neuromuscular diseases) (founder of the network which gathers 6 countries) (2009)
- CAIRE (Center of assistance and reintegration for women) in Guinea (2012)
- UNICEF, WHO as Consulting in the field of Health and Handicap.

AWARDS

- Knight of the National Order of Merit of Burkina Faso (2008)
- FITIMA, Better Guinea NGOs (2012)
- Winner of the Franco-German Prize for human rights in Guinea (2014)
- Exceptional leader discerned by the Collective of Guinean NGOs
THE REALITY OF RARE DISEASES IN WEST AFRICA

Rare diseases are a reality all over the world and African countries are not spared:

- Lack of information, screening and good diagnosis
- Economic precariousness

seriousness and the consequences of these pathologies
A - BARRIERS

- Socio-cultural burdens make patients:
  - hidden by their families
  - deprived of medical care, paramedical and educational system
  - abandoned, maltreated, stigmatized...
A - BARRIERS

- Weakness of the health system
- low number of hospitals and doctors
- inegalitarian system (between rich and poor)
A - BARRIERS

- Difficulties in accessing medicines
- Weakness of social security system
- Lack of information on rare diseases
- Failure to respect the fundamental rights to health
CONSEQUENCES OF THESE BARRIERS

the lives of hundreds thousands of patients with rare diseases and disabilities are in danger!
**FONDATION INTERNATIONALE TIERNO ET MARIAM (FITIMA)**

| Founded in 2003 | **Main Goal:** providing patients living with rare diseases and disabilities (*especially children*) with improved life | **Countries of intervention:** Burkina Faso and Guinea *ROAMY (West African network)* |
In this region

- Basic research and clinical research are rather marginal
- Few research centers that deal with large endemic areas (malaria, tuberculosis, etc.)

To fight against rare diseases, the emphasis is therefore on a system of care including: reception, diagnosis, paramedical care, adapted and inclusive education, social integration and information / awareness.
Occupational Therapy

Speech therapy

Hydrotherapy

Official launch of the training on "Women's Rights"
Access to patients

- Sensitization of the population and decision-makers using the media
- Allows patients to get out of marginalization and lead to behavioral changes

Key role of women
Improving diagnosis
"without proper diagnosis, no efficient treatment"

- Bring together local doctors and paramedicals specialists
- Organize multidisciplinary national medical consultations
- Define pathways of care
SETTING UP OF ROAMY

To go further in the best diagnosis, in sept 2009, FITIMA was the initiator of ROAMY (the west african neuromuscular diseases care network), which brings together practitioners from 5 countries (Burkina Faso, Guinea, Mali, Niger, Benin)

Sharing means, knowledges and experiences.
ROAMY

PLURIDISCIPLINARY CONSULTATION
OUAGADOUGOU - 2014
The ROAMY works in close collaboration with French specialists (especially from Hendaye and Cochin Hôspitals), in particular for:

- Making good samples in spite of the lack of dedicated laboratories
- Extracting DNA
- Refining diagnosis
- Providing medical and paramedical care
- Insertion into patient registers for possible clinical trials
- Establishing a genuine international collaboration
ROAMY

A CASE OF DISFERLINOPATHY

DUCHENNE NEUROMUSCULAR DISEASE
TRAINING

- Participation of 10 caregivers either in advanced training (in France)
- International scientific conferences
- Publication of articles and presentation of posters
INTRODUCTION

Myopathies are neurological rare pathologies, characterized by primary involvement of muscle tissue damage. The clinical presentation is progressive muscle weakness, myalgia and voluntary muscle atrophy. Studies on myopathies are nonexistent; this study is the first with genetical results.

METHODS

Descriptive, retrospective and analytical, study of 10 years (2004-2014). We reported 17 cases (12 male, 5 female) from 16 families followed up in neurology in Thierno and Mariam International hospital of patients were 19.83 years, from 5 to 30 years. They were followed up for a mean time of 4.29 years.

RESULTS

The mean age of the family is 25.5 years. The mean age of the patient is 19.8 years. The most affected muscle group is the quadriceps femoris, followed by the gluteus maximus, adductor magnus, and tibialis anterior. The most common signs and symptoms are progressive muscle weakness, myalgia, and muscle atrophy.
PUBLICATION IN SCIENTIFIC REVIEW

MYOLOGIE DANS LE MONDE

ROAMY : l’expérience de quatre pays d'un jeune réseau d'Afrique de l'Ouest

MADAGASCAR : un terrain d’expérimentation

Le ROAMY, Réseau Ouest Africain de prise en charge des Myopathies, a vu le jour en septembre 2009. Il rassemble les professionnels concernés du pays d’Afrique de l’Ouest ainsi que de la France. Des maladies neurovasculaires sont en effet présentes sur le continent africain avec des spécificités de Duchenne. Ce réseau permet une meilleure prise en compte de ces pathologies. Cependant, des efforts sont encore à faire pour sensibiliser le diagnostic et pour aller vers une prise en charge multidisciplinaire.

Réseaux de ROAMY

À l’Afrique du Sud, en Afrique de l’Est et en Afrique de l’Ouest, ces derniers ont mis en œuvre des projets visant à former les médecins pediatriques et neurologues, à sensibiliser les médecins aux différents aspects de la maladie et à améliorer la prise en charge des patients.

Revue de ROAMY

À Madagascar, le thème de la neurologie a été retenu. En effet, les maladies neurovasculaires sont plus fréquentes en Afrique. Il est donc impératif de former les médecins à ce sujet. Le ROAMY a donc mis en place des ateliers et des formations pour sensibiliser les médecins à ces maladies.

Conclusion

Ce projet ROAMY est donc une belle initiative qui permet de sensibiliser les médecins à la prise en charge des maladies neurovasculaires. Il est important de continuer ces efforts pour améliorer la prise en charge des patients.

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C – HOPE TO ACHIEVE (NEW COLLABORATIONS)

❖ To Improve diagnosis and Access to treatment:

❖ Develop ROAMY
❖ Enable the establishment of patient registries and DNA libraries
❖ Establishment of a sub-regional laboratory dedicated to rare diseases

❖ Training of therapists
❖ Access to Treatment
❖ Financial assistance
❖ Infrastructure for the taking care of patients
CONCLUSION

- We see that needs are important but intelligent and efficient collaboration between patients (whose rights to health is denied), governments (which still need to be sensitized) and industrialists (who can find real niches) could allow hundreds of thousands of patients to be cared for.

- It is also known that any success, any discovery on a so-called rare disease can be very useful for more common diseases.
THANK YOU VERY MUCH FOR YOUR ATTENTION!

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