## Appendix 1. Advisory board

### Advisory board participants

| Name | Country | Comment |
|---|---|---|
| Dr Mohammed Al Jumah | KSA | Advisor |
| Dr Mohammad Al Muhaizea | KSA | Advisor |
| Dr Ahmed Al Rumayan | KSA | Advisor |
| Dr Abdulaziz Al Saman | KSA | Advisor |
| Dr Ali Al Shehri | KSA | Advisor |
| Dr Edward Cupler | KSA | Moderator |
| Dr Samah Ishak | KSA | Advisor |
| Dr Mohammed Jan | KSA | Advisor |
| Dr Ayaz Shah | KSA | Advisor |
| Dr Abubaker Al Madani | UAE | Advisor |
| Dr Wassim Fathallah | UAE | Advisor |
| Dr Pawan Kashyape | UAE | Advisor |
| Dr Gururaj Kodavooru | UAE | Advisor |
| Dr Cristina Skrypnyk | BAHRAIN | Advisor |
| Dr Gholamreza Zamani | IRAN | Advisor |
| Dr Laila Bastaki | KUWAIT | Advisor |
| Dr Andre Megarbane | LEBANON | Advisor |
| Dr Khalid Elthilily | OMAN | Advisor |
| Dr Sylvie Tuffery Giraud | FRANCE | Advisor |
| Dr Andoni Urtizberea | FRANCE | Advisor |
| Dr Carlos Ortez | SPAIN | Advisor |

## Appendix 2. Questionnaire

### Discussion guide for MENA DMD expert meeting – 23 September, 2016

*Facilities/Organization*

Describe the **type of clinics** where DMD patients are mostly diagnosed and/or followed-up

- Public/private ☐
- Dedicated neuromuscular clinic ☐
- Multidisciplinary clinic (if any) ☐

### Referral process

1. What is the main source of DMD patients referral in your accounts?

a. Pediatricians, child neurologists ☐
b. Neurologists ☐
c. Geneticists ☐
d. Family medicine ☐
e. Other specialties ☐
f. Teachers, relatives ☐

2. What are the motives of the DMD patient referral?

a. Muscle of weakness ☐
b. Isolated elevated of serum CK levels ☐
c. Walking delay/difficulty ☐
d. Frequent falling ☐

e. Genetic counseling/family history ☐
f. Other complications (e.g., cardiac, respiratory) ☐

### Number of DMD patients

- Approximate caseload of patients with muscular dystrophy and among them cases of DMD? .....
- Number of DMD patients with confirmed point mutation?.....
- Number of DMD patients with pending sequencing of the DMD gene? .....
- Number of DMD patients without MLPA or NGS? .....
- Approximate proportion of non-Nationals (from inside or outside the country) with DMD? .....

### Technical facitilies:

In your clinic do you have access to:

- DMD PCR genetic studies – locally ☐ sent abroad ☐
- DMD MLPA genetic studies – locally ☐ sent abroad ☐
- DMD mutation specific genetic studies - locally ☐ sent abroad ☐
- Muscle biopsy ☐
- Respiratory investigations (FVC, others) ☐
- Cardiac investigations (echocardiography) ☐
- PGD (preimplantation genetic diagnosis) ☐

### Staff

Do you have in your team a dedicated MD to deal with DMD patients/neuromuscular patients

Yes ☐ No ☐

Is a physiotherapist or other paramedic assisting you in the clinic?

Yes ☐ No ☐

Would you like to support training for your physiotherapist in terms of managing DMD patients?

Yes ☐ No ☐

### Timeline

Average time needed on average to reach a final diagnosis of DMD:

- Time between first symptoms and going to the physician
- Time between first referral and identification of a deletion/duplication in the DMD gene
- Time between first referral and identification of a point mutation in the DMD gene

## *Diagnosis*

1. What are the current diagnostic steps for DMD patients in your countries?.....

2. What is the diagnostic algorithm for DMD patients in your countries?.....

3. Considering the availability of several means of DMD diagnosis at your disposal, which one would you adopt as a standard:

a. Muscle Biopsy
b. Multiplex PCR
c. MLPA test
d. MLPA test followed by sequencing of the DMD gene (if MLPA negative)
e. Targeted exome sequencing – panel of genes (i.e, muscular dystrophies); What action would you take typically if negative result found?
f. Whole exome sequencing

4. How could you/we improve the process of early point mutation defection in DMD patients?

- Is it a financial issue for you and/or the patients and relatives? Yes ☐ No ☐
- Do you need a regional center for DMD sequencing? Yes ☐ No ☐

- Would you suggest that we should screen all DMD patients with sequencing prior to any MLPA test?

  Yes ☐ No ☐

- Would you like send your patients DNA abroad still for sequencing?

  Yes ☐ No ☐

  Do you envisage any obstacles with sending Dried Blood Spot (DBS) test samples abroad for sequencing?

## *Data collection – registries*

5. Do you collect the genetic and clinical information either formally or informally currently in some kind of repository? Yes ☐ No ☐

6. How important would it be to collect this information in a registry to support patient follow-up and publications? Yes ☐ No ☐

7. Do you feed (or would like to do so) registries for DMD patients

- At the national level Yes ☐ No ☐
- At the regional level (GCC) Yes ☐ No ☐
- At the international level (TREAT-NMD or others) Yes ☐ No ☐

## *Natural history*

8. What is the typical natural history for diagnosed DMD patients in the Middle East region, once a differential diagnosis has been made?.....

9. What is the typical life expectancy of DMD patients in the Middle East?.....

10. At what age do DMD patients typically become nonambulant on average (100% wheelchair use)?.....

11. What's the proportion of DMD patients being on corticosteroids? .....%

12. Do you feel these DMD patient management parameters have improved over the past 20 years in your country?

Yes ☐ No ☐

## *Follow-up – outcome measures*

13. Which clinical and patient-reported outcome measures are available in your clinic;

At ambulatory stage:

- Only symptoms and disabilities reported by patient and relatives Yes ☐ No ☐
- Manual muscle testing (MMT) Yes ☐ No ☐
- 6-min walking test (6MWT) Yes ☐ No ☐
- Other timed function tests Yes ☐ No ☐
- Functional scales (North Star Scale, MFM, others) Yes ☐ No ☐
- Myotools Yes ☐ No ☐
- Other (quality of care scales) Yes ☐ No ☐

At nonambulatory stage:

- Same as above Yes ☐ No ☐
- Respiratory and/or cardiac outcome measures? Yes ☐ No ☐

14. Which criteria do you use to define whether a DMD patient is nonambulatory relative to an ambulatory patient?.....

15. What are your experiences and feelings about the 6MWT?.....

16. How valid is 6MWT test as a standard outcome measure of DMD disease progression?

a. In your clinical practice?

b. In clinical research studies?.....

17. To what extent do you follow the international guidelines (standards of care)?

- Published by TREAT-MND or others Yes ☐ No ☐ Partially ☐
- To what extent would it be helpful to have such guidelines adapted for our colleagues in the Middle-East region?

18. How many of your nonambulatory patients are on

- Noninvasive ventilation? .....
- Tracheotomy? .....

19.How many of them underwent spine surgery? .....

20. How many of them travel abroad:

- For second opinion in an expert neuromuscular center (and where precisely?) .....
- For nonconventional therapies (stem cell therapies, others)? .....

## *Ataluren therapy*

1. What is your current management approach for treatment of nonsense mutation DMD patients?

a. Corticosteroids only
b. Ataluren only
c. Ataluren + corticosteroids

2. What would your optimal management approach for nonsense mutation DMD patients?.....

3. What are your treatment expectations regarding ataluren? (efficacy)

a. Slowing disease progression
b. Stabilization of symptoms
c. Complete recovery
d. Depends on the stage of the disease when treatment initiated, relative to natural history

4. Did you notice any side effect in your patients on Ataluren (safety)?; Would they prompt you to discontinue the treatment? Yes ☐ No ☐

5. How easy is it for you and your nonsense mutation patients to access ataluren treatment in your account and what could be done to accelerate and facilitate access?.....

6. What are the major obstacles? Diagnosis, reimbursement or other?.....

7. Considering that one of the key treatment objectives in DMD is to maintain muscle function,

a. How would you address a patient who is nonambulatory, but has upper limb function?.....
b. How would manage a patient on treatment that recently lost ambulation while on therapy?.....

8. If any, which your nonsense mutation DMD patients would you consider not treating with ataluren? Why?.....

## *Patient support – awareness – education*

21. What is required in terms of a patient awareness program or patient support program?

a. Patient awareness sessions and materials
b. Assigned nurse education
c. School/kindergarten/sport teacher awareness programs
d. Educational awareness sessions for different physician specialties
e. Website, facebook or other social media

22. Is there a patient support group/organization in your country? Yes ☐ No ☐

- If so, is it dedicated to neuromuscular patients exclusively? Yes ☐ No ☐
- Or to rare diseases Yes ☐ No ☐
- Or to disabled people as a whole Yes ☐ No ☐

### *Prospects*

How do you feel **identification, management** and **treatment** of DMD patients can be improved in the Middle East?.....

What recommendations and tools would you propose for improving long-term outcomes in DMD patients in your country?.....