Scientific Advisory Board
Fragile X

AFrAHM asbl
Monday June 3th, 2013

AFrAHM a.s.b.l
X fragile

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Agenda

- Governance
- Fragile X Syndrome
  - Objective 1: promote research & clinical trials
  - Objective 2: inform Belgian families: conference 2013/14
  - Objective 3: increase awareness of Fragile X with Belgian paediatricians
- Associated disorders (premutation, FXTAS, FXPOI)
  - Objective 4: share a clear position on associated disorders

Governance

- Who’s who ?
- Roles & responsibilities
  - AFrAHM & Association XFB: Parent’s support, information hub, education projects, social events, awareness campaign, promote research & clinical trials
  - SAB: advise the association on medical matters, promote research & clinical trials
- Action Plan XFB
- European Fragile X Network & other associations
- Frequency of our meetings (2x per year ?)

Objective 1

Promote research & clinical trials

Objective 1: promote research & clinical trials (1/2)

- Basic research: latest results? What to expect?
  - mGluR5 antagonists
  - GABA-B agonist
  - Minocycline
  - Dendritic spines (FRAX486),…
- FRAXA grants
  - Also in Belgium (first int’l partner outside Americas)
  - http://fraxa.org/applyinggrants.aspx

Objective 1: promote research & clinical trials (2/2)

- Clinical trials
  - Worldwide
  - Belgium: When? Where? Target population? Comms?
- Fragile X Clinical & Research Consortium
  - www.fxcrc.org
  - Looking for international affiliates

Objective 2

Inform Belgian families : conference 2013/14

Objective 2: inform Belgian families: conference 2013/14

- Fragile X conference in Belgium in 2013/14 – what are our options? Main topic?
- Attendance at NFXF conference July 2014 (Anaheim, Ca, US)?
- How to best summarize latest research to families?
- Scientific Advisory Board (SAB) to review/validate our website (www.x-fragile.be)
  - Can we publish your summary details on SAB webpage?

Objective 3

Increase Fragile X awareness with Belgian paediatricians

Objective 3: increase Fragile X awareness with Belgian paediatricians

- Fragile X presentation @ Groupement des Pédiatres Francophones Belges (GPFB) – October 2013
  - After the presentation of XFB and the services we can offer, we would like a doctor (member of the SAB?) to present the key medical facts

- Add a Fragile X screening questionnaire to our website (www/x-fragile.be)
  - Fryns & Borghraef (2001) or is there anything more recent?
  - How to get legal rights to use on our website
  - Could we have a version for use by doctors and a simplified one for parents?

- Postcards sent to all paediatrician – building on the NFXF (US) success story
  - We would like the SAB to sign-off our message

Example of Postcard

Who's affected:
Approximately one in 3600 males and one in 4000 to 6000 females are affected. One in 300 women and one in 800 men are carriers.

All children with significant developmental delay of unknown origin should be tested for fragile X.

What to look for:
Physical symptoms, in post-pubertal males, can include a long, narrow face, prominent ears and enlarged testicles—however, lack of physical features does not rule out fragile X.

Some female carriers experience early menopause (POF) and some male carriers develop neurological difficulties later in life (FXTAS).

What can be done:
By ordering a *FMR1-DNA blood test*, this disorder can be ruled out. The test, which is better than 99% accurate, is usually covered by insurance, including Medicaid.

Many individuals are undiagnosed. A diagnosis can open up many doors to services and support.

Where to find help:
Phone the National Fragile X Foundation for more information, or visit our website to view over 1200 pages of content *including detailed testing information.*

1-800-688-8765 or **www.FragileX.org**

Please join us at our 9th International Fragile X Conference in Washington DC, June 23–27, 2004.

The National **Fragile X** Foundation
P.O. Box 190488
San Francisco, CA 94119-0488

What do pediatricians need to know about

fragile X syndrome?

It's the **world's leading cause** of inherited mental impairment, ranging from mild learning and psychological problems to autism and severe mental retardation.

Objective 4

Share a clear position on associated disorders

Objective 4: share a clear position on associated disorders

- Premutation
  - Information to (extended) families
  - Information to teenagers (<18 years old)
    - Early pregnancy risk & associated disorders
- FXTAS / FXPOI
  - Communication to patients

Thank you!

Your personal commitment makes the difference and gives hope to families

Association X fragile Belgique

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  www.x-fragile.be
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  https://www.facebook.com/AssociationXFragileBelgique
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