

## FSG Annual General Meeting August 2008

### Guest Speaker: Dr Wilcox

The 2008 Fabry Support Group AGM, was held in August at the Stamford Plaza, Mascot Sydney, was well attended by members of the group and guests.

We were very fortunate this year to have an international guest speaker from USA to present on the day. Our thanks go to Dr Wilcox, who was in Australia to visit doctors and professionals who treat people with Fabry disease and to Genzyme for organising and sponsoring the event.



#### **William Wilcox, MD, PhD**

Director, Inborn Errors of Metabolism; Director, Skeletal Dysplasia Morphology Lab

William R. Wilcox, MD, PhD is Director of the Metabolic Disorders Clinic in Medical Genetics and Director of the Skeletal Dysplasia Morphology Laboratory at Cedars-Sinai. Dr. Wilcox also serves Cedars-Sinai as Director of the Lysosomal Storage Disease Treatment and Research Centre, Residency Director for the Medical Genetics Residency and Assistant Professor of the Medical Genetics Fellowship Program.

Dr. Wilcox is board certified in paediatrics, clinical genetics, clinical biochemical genetics and clinical molecular genetics. His current research interests include enzyme replacement in lysosomal storage diseases; the morphology, genetics and pathophysiology of human dwarfing conditions (skeletal dysplasias); and identification of the genetic defect in the Marinesco-Sjögren syndrome. He is on the advisory board of the Fabry Disease Registry and the Genetic Leadership Collaborative.

Dr. Wilcox is a graduate of the University of California, Los Angeles (UCLA) School of Medicine. He completed a residency in paediatrics at UCLA and a genetics fellowship in the UCLA Intercampus Medical Genetics Training Program.

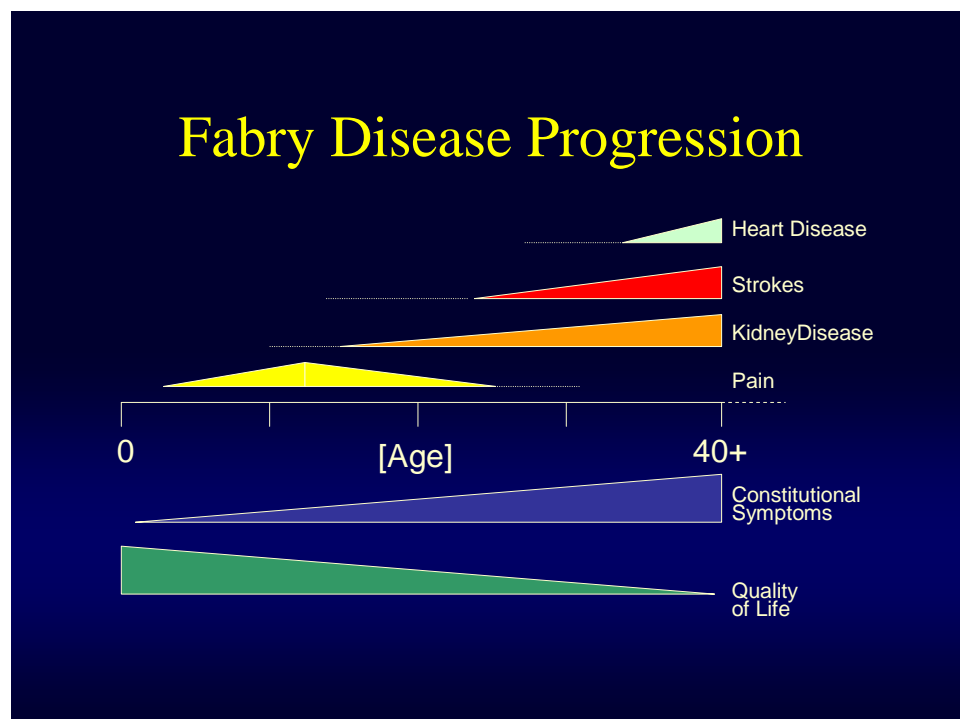


Dr Wilcox presented an informative and interesting overview of the 'Management of Fabry Disease in 2008 and Beyond'. Topics included the clinical manifestations of the disease, progression, diagnosis, family screening, issues for females and children, enzyme replacement therapy, and the international Fabry Registry Program. In closing Dr Wilcox gave a comprehensive explanation of Chaperone Therapy which is still in clinical trials.

Some of the key points that were mentioned in Dr Wilcox's presentation include:

- The term "female carriers" should be left in the past – two thirds of women with Fabry disease will have significant symptoms at some stage of their life
- Many women deny their illness however problems in women are often 'silent' – it is imperative to have regular assessments to detect problems early
- Children are treated much earlier in America – and girls can have just as many symptoms as boys
- Teenagers can have significant disease symptoms so early diagnosis, assessment and monitoring is important
- Treatment guidelines in America are generally much earlier than Australia – we wait too long.
- If you have Fabry disease and are monitored by a specialist you still may not be on the international Fabry Registry Program unless you are receiving ERT...ask your specialist next time you see them. Children in Australia are not on the International Fabry Registry Program.

Dr Wilcox has kindly given permission to reproduce parts of his presentation for the benefit of FSG members who were unable to attend the meeting. Interested FSG members can also request a full copy of the presentation by emailing the FSG President, Megan Fookes. The presentation is full of thorough and current information and is worth a look. If you do request a copy please keep in mind some of the information (e.g. guidelines for starting ERT) will be specific to the U.S.A.



## Fabry Family Screening

- Family screening extremely important
  - inherited disorder
  - new mutations infrequent
- One diagnosis can uncover many others
- Early diagnosis and intervention can lead to improved outcomes
- Important for families to understand risk of passing disease gene

## Female “Carriers”

- From multiple studies, approximately 2/3 of women will have significant symptoms at some point in their life
- Significant kidney disease occurs in about 1/3, but kidney failure is less common than with men (about 10% of women)
- Heart disease and strokes are the most common serious problems. The onset is about 10 years later than men.
- Exercise intolerance, fatigue, pain, temperature intolerance or lack of sensation, intestinal and stomach problems, anxiety, depression, and headaches can be debilitating
- There is currently no way to predict which women will have serious involvement later in life
- Most doctors think women can't have symptoms
- Many women deny their illness

## Children with Fabry

- Boys begin having problems with pain at an average age of 5
- Problems with sweating (overheating) and exercise are common in school
- Stomach pain and diarrhea are common
- Teenagers can have heart disease, strokes, and significant kidney disease
- Girls can have as many problems as boys

## Fabry Registry Program

- World-wide program
  - Follow patients over time- what problems does enzyme prevent, what doesn't it help
  - Allows your results to be tabulated and tracked easily over time
  - Data is entered with a code
  - Data from your records can be entered even if you are not seen at the center- with your consent
  - PLEASE PARTICIPATE!
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