

Rare Disorders Denmark

# Patient Involvement and National Standards of care – The Example of Tuberous Sclerosis

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## Outline

- Danish TSC Association
- What is TSC?
- Why are standards of diagnosis and care important?
- TSC guidelines
- How were they developed?
- Patient organisation involvement





## Danish TSC Association

- Founded in 1992
- Working to improve knowledge and treatment of Tuberous Sclerosis Complex
- Board members are all volunteers- mostly parents
- Around 140 family memberships today
- I have been chairman since 1999





## What is TSC?

- Rare genetic disorder
- Form cancerous -like benign tumors throughout the body in multiple organs -brain-kidneys-lungs
- Often causes developmental disturbance and mental retardation leading to challenging behaviour
- Prevalence of 1:7000



## Why are standards of diagnosis and care important?

- Important due to the rarity – general physician has little knowledge
- Important to collect knowledge/experience, globally, in clinical guidelines
- Should also include the psychiatric/psychological aspects of the disease



## Why are standards of diagnosis and care important?

- Especially important for diseases that affects many organs
- They require treatment of many different conditions





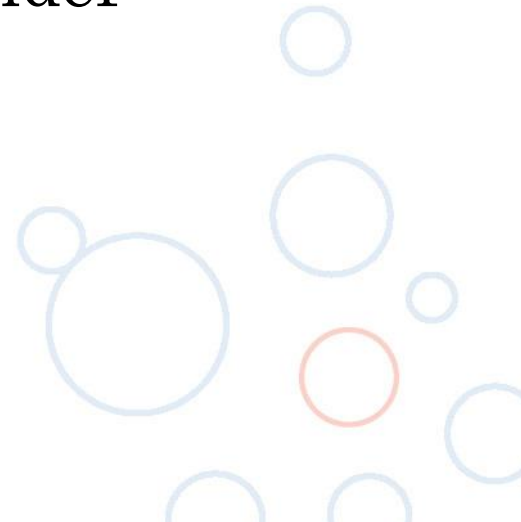
# TSC guidelines and how they were developed

- In the present form the global TSC guidelines mainly contains information on the clinical aspects like
- the criteria for diagnosis
- How to treat and monitor the clinical aspects (epilepsy, brain and kidney scans)



# TSC guidelines and how they were developed

- Only very briefly touches how the cognitive and behavioural problems can be monitored and relieved
- One of the reasons why they are under revision







## TSC guidelines and how they were developed

- The global guidelines was developed in collaboration between
  - Clinicians
  - Researchers (geneticists, neurologists, kidney experts, psychiatrists, psychologists etc)
  - Patient organisations (mainly from UK and USA but PO from other countries were also involved)and their experiences
  - First edition was published in 2001



## TSC guidelines and patient organisation involvement

- The Danish guidelines is a translation of the 2001 version but include more information on the cognitive and behavioural aspects
- The guidelines was adjusted to the Danish Healthcare system
- Collaboration between professional doctors and psychologists and the Danish TSC association to ensure the validity of content



## TSC guidelines and patient organisation involvement

- The translation was made on the initiative of the Danish TSC association
- They are implemented at one of the centres for Rare diseases in Denmark but only for children
- There is no medical centre where adults with TSC are followed. They are send to neurologists, kidney specialists etc. that are not experts in TSC and do not coordinate their treatment



## TSC guidelines and patient organisation involvement

- The global TSC guidelines is presently being updated including new developments as well as the psychiatric and psychological aspects
- This is conducted in close collaboration with clinicians, researchers and PO's
- Hence, an evaluation of the former guidelines



## TSC guidelines and patient organisation involvement

- Danish TSC must revise the guideline as well
- We are working on implementing the guidelines for adults by participating in developing the plan for the National Strategy on Rare Diseases in Denmark
- Alone we are too small-together we are strong 😊