



MITOCHONDRIAL
MEDICINE SOCIETY

MMS UPDATES for MitoAction

April 1, 2016

New Projects

- 1) Solid Organ Transplantation: spearheaded by Dr Parikh
- 2) Stroke protocol for MELAS publication: spearheaded by Dr Koenig
- 3) Standards of Care for Mitochondrial Disease Patients
- 4) Centers of Excellence

Solid Organ Transplantation

- Poster being presented by Dr Parikh at SIMD 2016
- 35 patients, 17 mito centers, 4 countries (US, Canada, UK, Australia) receiving heart, liver or kidney transplant
- Half the patients had transplant before diagnosis was known
- Overall survival was similar to those in transplant literature with exception of POLG-related disorders and liver transplant

Review | March 07, 2016

**Recommendations for the
Management of Strokelike
Episodes in Patients With
Mitochondrial
Encephalomyopathy, Lactic
Acidosis, and Strokelike Episodes
ONLINE FIRST**

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JAMA Neurol. Published online March 07,
2016. doi:10.1001/jamaneurol.2015.5072

Preventative Care Screening Guidelines

1. Anesthesia
2. Audiology
3. Cardiac
4. Critical Illness
5. Endocrine
6. GI
7. Hematologic
8. Immunologic
9. – 12. Neurologic 1 – 4 (development, headaches, stroke, spasticity, seizures)
13. Ophthalmologic
14. Orthopedic
15. Pregnancy
16. Psychiatric
17. Pulmonology/Respiratory
18. Renal, Acid/Base, Electrolytes
19. Surgery precautions
20. Constitutional (Fatigue, Pain)

Preventative Care Screening Guidelines Subjects

- 1. Anesthesia** (no subgroup needed; work completed)
 - a. Will include recommendations from Consensus Statement (Genetics in Medicine Dec 2014)

- 2. Audiology**
 - a. SNHL
 - b. Cochlear implants

- 3. Cardiac**
 - a. Cardiomyopathy
 - b. Dysrhythmias
 - c. Pacer placement (especially for Pearson/KSS)
 - d. Hypertension
 - e. Chest pain

4. Critical Illness

- a. Endocrine worsening (especially adrenal insufficiency)
- b. Acid/base status
- c. Worsening RTA
- d. Dysrhythmia
- e. Respiratory issues related to weakness
- f. Other?

5. Endocrine

- a. Thyroid
- b. PTH/Ca/Vit D
- c. Adrenal
- d. Diabetes
- e. Short stature/GH deficiency
- f. Bone density (related to disuse/non-ambulatory status)

6. GI

- a. Liver disease
- b. Constipation
- c. Motility
- d. Nutrition
- e. FTT

7. **Hematologic**
 - a. Anemia
 - b. Bone marrow failure

8. **Immunologic**
9. **Neurologic 1**
 - a. Epilepsy*
 - b. Headache/Migraine
 - c. Movement DO
10. **Neuro 2**
 - a. Myopathy
 - b. Neuropathy*
 - c. Tone
 - d. Spasticity
 - e. Dystonia
11. **Neuro 3**
 - a. Developmental Delays
 - b. Learning Disabilities
12. **Neuro 4**
 - a. Stroke – refer to published guideline
13. **Ophthalmologic***
 - a. Retinal and optic nerve disease
 - b. Ptosis
 - c. Ophthalmoplegia

- 14. **Orthopedic**
 - a. Scoliosis
- 15. **Pregnancy***
- 16. **Psychiatric**
 - a. Depression
 - b. Anxiety
 - c. Personality disorder
- 17. **Pulmonology/Respiratory***
 - a. Central and obstructive apnea
 - b. Respiratory insufficiency
 - c. Sleep (include central/CNS related issues)
- 18. **Renal, Acid/Base, Electrolytes**
 - a. Acidosis
 - b. RTA
 - c. Glomerulonephritis
- 19. **Surgery precautions** (no subgroup needed; work completed)
 - a. Refer to consensus statement
- 20. **Constitutional**
 - a. Fatigue
 - b. Pain
 - c. Weight management
 - d. High altitude travel

Creating Mitochondrial Centers of Excellence

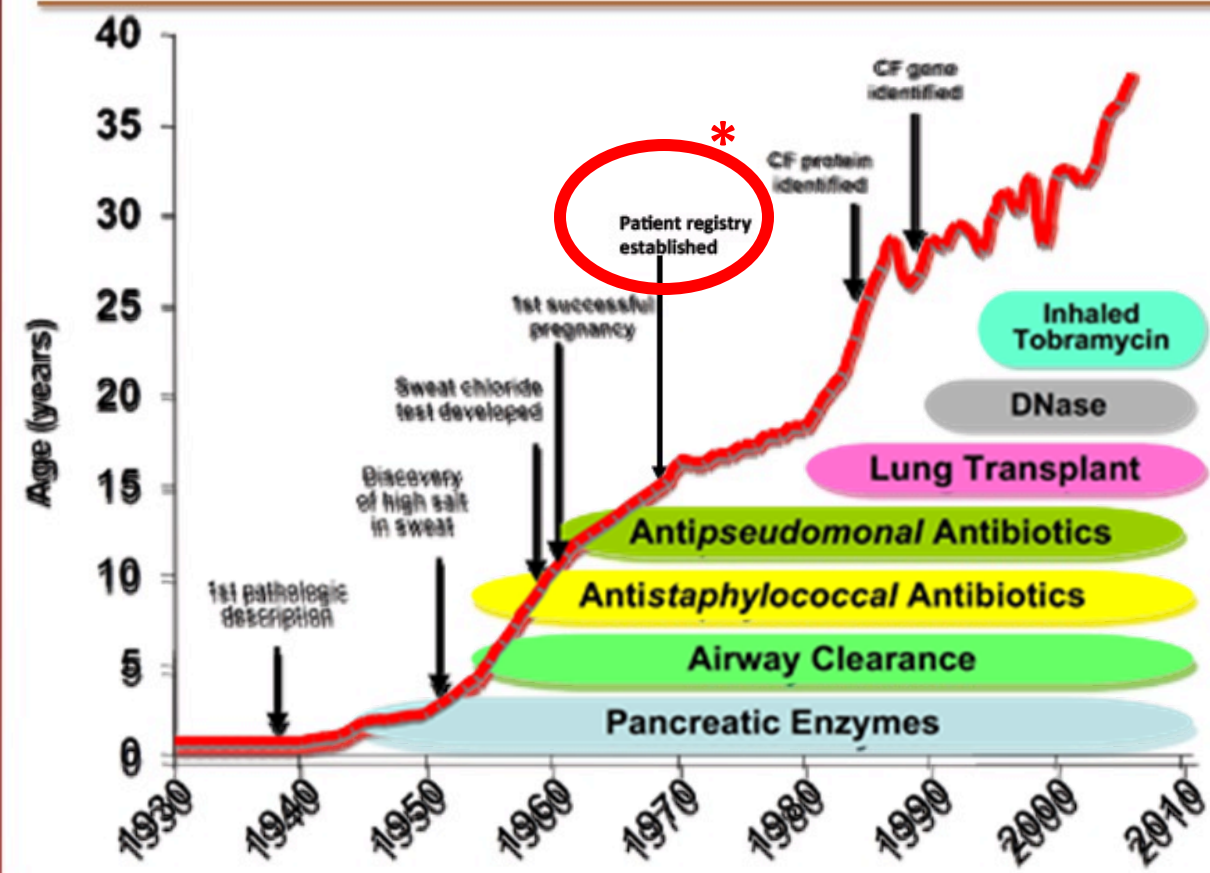
- 1) Focus groups of patients, families, caregivers
- 2) Input from Support Groups (MitoAction, UMDF, LHON)
- 3) Creation of Surveys for further data collection
- 4) Distribution of Surveys and Data Collection
- 5) Create checklist of COE requirements
- 6) Checklist questionnaires collected from potential COE
- 7) Governing Board to check COE is following Standards of Care and fulfilling requirements

We will be seeking out the community's help to establish what a Mitochondrial COE should look like; stayed tuned!

Survival in CF – an incurable genetic disease

- Cystic fibrosis remains “incurable”
 - Complications involve:
 - Lung
 - GI tract
 - Pancreas
 - Kidney
 - Bone
 - Mood/behavior
- ...yet life expectancy has increased 10-fold in the past 60 years

CF: “symptomatic therapy” improves median survival



	simple	cheap	pre-exist	non-pharm
Inhaled Tobramycin	✓			
DNase			✓	
Lung Transplant				
Antipseudomonal Antibiotics	✓	✓	✓	
Antistaphylococcal Antibiotics	✓	✓	✓	
Airway Clearance	✓	✓	✓	✓
Pancreatic Enzymes	✓	✓	✓	

Source: www.nationaljewish.org

*Development of Patient registry, CF Centers of Excellence and Standardized Care help improve overall morbidity and mortality for CF patients ; addition of standardized treatments at every center that are: Simple, Cheap, Pre-exist, and Non-pharmacological