

RETT SYNDROME IN GREECE

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Maria...19yo



Passed away...1-2010
Respiratory infection...C-R arrest

Angels on earth



9-2011 Gave birth to the:
Rett Syndrome Association Of Greece

Site: www.rettgreece.gr

2011: Population: 11,000,000, only 106,428 live births



50%(106,428) of newborns = 53,000 girls

Incidence: 1:10,000
~ 5 new cases/year
Prevalance ~ 100 girls for last 20 years



Present status

3 Pts Dr Evangeliou

21 Pts: Dr zafiriou

Dr Tzoufi ?

27 Pts Dr Par/kos

50 MECP2+
Dr Frysira

Dr Vorgia



Rett syndrome checklist

Illinois Rett syndrome association

RETT SYNDROME HEALTH CARE GUIDELINES

Name: _____ Birthday: _____

Age	9	10	11	12	13	14	15	16
Clinical Diagnosis								
Molecular Diagnosis								
Audiological Evaluation*								
Ophthalmologic Evaluation*								
Seizures								
Health Maintenance								
Weight for Height percentiles								
Head circumference								
Oral-ocular function								
Bruxism/Dental								
Scoliosis check								
Hyperventilation/apnea								
Aerophagia								
Constipation								
Abdominal pain/gall bladder								
Tanner stage								
Range of motion								
Pathologic fx/lane density								
EKG								
Neurologic status - Muscle bulk								
Muscle tone								
DTRs								
Ataxia								
Developmental status								
Ambulation								
Communication								
Temperament/Mood								
School Program								
Therapies								
Family Functioning								
Medications								
Allergies								

* Document once. If normal, repeat only as needed

*For inferential purposes only

Rett care:

- Apply the Illinois RSA guide
- Define problems and address them
- Notably: epilepsy(VPA & LTG)
- Orthopedics, endocrine
- Respiratory abnormalities
- Nutrition, constipation

Rett care:

- Re-evaluation every 6 months or sooner if necessary

Rett experts

- We have none
- We learn from the patient and parents

<< Expertise center >>

One man show: It is primarily **volunteer** work by **few professionals** who have the desire to give of themselves for the children and families

Most of the work is being done by ***Maria's mother*** and few other parents

Hx of the parent organization:

- Created in 9-2011
- Functions under the auspices of the Hellenic union of rare disorders

Goals:

- To promote education to
professionals and families
- To improve care for the children
- To ascertain that families procure
entitled benefits
- Provide psychosocial support families

Activities and projects

- To evaluate and place the children in a regular follow-up care program
- Participates in scientific meetings, both medically and as a parent group
- Organizes outings, a marathon, bazaar,
- >> parties for children and families
- Provides support for whatever needs the families have: psychosocial and economic

Wishes and Aims

- Ideal to create a registry and enlist all the children with the syndrome in Greece
- To overcome problems such as where to hospitalize small weight adults
- Better care with associated sxs as PT, OT, speech, swallowing studies etc
- Ideally to staff a Rett expertise group in Athens and one in northern Greece

Thank you!