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 Geece

Site: www.rettgreece.gr

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

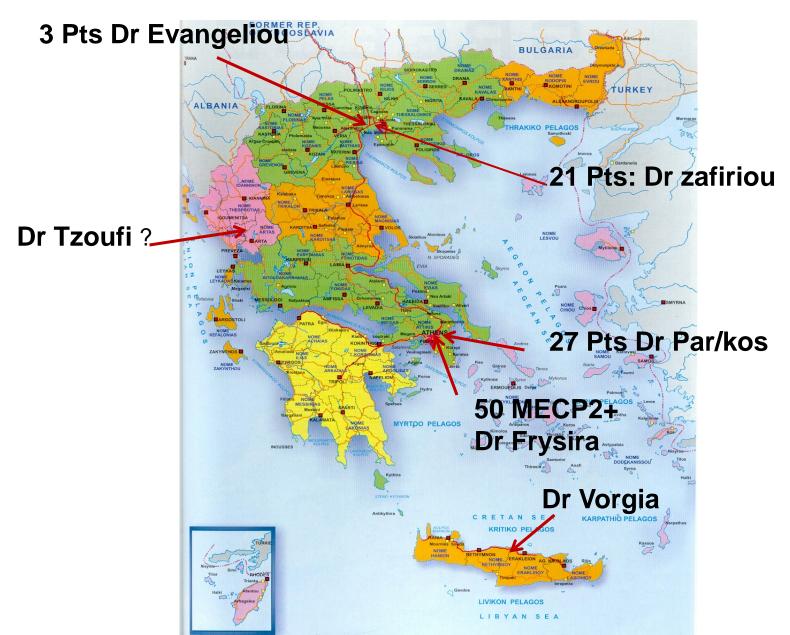


50%(106,428) of newborns = 53,000 girls

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



Present status



Rett syndrome checklist Illinois Rett syndrome association

RETT SYNDROME HEALTH CARE GUIDELINES

Age	9	10	2.1	12	13	14	15	16
Clinical Diagnosis								
Molecular Diagnusis								
Audiological Evaluation								
Ophthalmologic Evaluation'								
Seizurcs			Γ					
Health Muintenance								
Weight for Height percentile			L					
Head dreumference				1				
Orsi-mater function	l							
Bruxism/Dental								
Scolinsis check								
Hyperventilation/apnea								
Aerophagia		·						
Constigation								
Abdominal pain/guil bludder								
Tanner stage								
Range of motion								-
Pathologic fx/Bone density								
ICKG		L	L					
Neurologic status - Muscle bulk								
Muscle tone								
DTRs								
Ataxis								
Developmental status								
Ambulation			1					
Communication								
Temperament/Mond	1							
School Program							i	
Therapies					1			
Furnity Functioning								
Mediculians				-				
Allergies								

^{&#}x27;Docqueent once. If normal, repeat only as needed

*For informationed purposes only

2002

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, bazzaar,
- >> 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

