National Based, Retrospective Study on the Evaluation of Clinical, Laboratory, and Imaging Research of Angelman- Turkey's Multicenter Study

*Hakan Gümüş, *Ayten Güleç , *Mehmet Canpolat, Betül Kılıç, Özlem Yılmaz, Yasemin Topçu, Seda Kanmaz, Adnan Deniz, Ünal Akça, Gülten Öztürk, Muhammed Bahadır Samur, Peren Perk Yücel, Dilara Füsun İçağasıoğlu, Aydan Değerliyurt, Mesut Güngör, Hülya İnce, Pınar Özkan, Kürşad Aydın, Sanem Yılmaz, Hasan Tekgül, Selcan Öztürk, Bülent Kara, Burcu Karakayalı, Gülen Gül Mert, Nihal Olgaç Dündar Dündar, Çisil Çerci Kubur, Sedef Terzioglu Öztürk, Pınar Gençpınar, Aslıhan Kıraz, Murat Erdoğan, Munis Dündar, *Hüseyin Per, Turkish Pediatric Angelman Syndrome Study Group

*Erciyes University Faculty of Medicine, Department of Pediatric Neurology/Turkey

ICNC 2022
17th INTERNATIONAL CHILD NEUROLOGY CONGRESS
ANTALYA, TURKEY I OCTOBER 3-7, 2022

INTRODUCTİON

Angelman syndrome (AS) is a rare genetic neurodevelopmental disorder with severe learning difficulties, ataxia, and dysmorphic facial features. It often has a characteristic epileptic EEG pattern. They are happy and social patients. disturbance There are often jerky movements, frequent and sometimes inappropriate laughter, water addiction, and sleep.

OBJECTİVES

It is an important genetic disorder that should be considered in behavior, language, and gait disorders that accompany a happy and sociable personality. A comprehensive evaluation was aimed in terms of epilepsy frequency and treatment approaches.

MATERİALS& METHODS

Patients diagnosed with Angelman syndrome between 01.01.2010 and 01.01.2022 through a 14-centered retrospective study in Turkey were followed up for at least 3 months. Their presenting complaints, demographic data, radiological findings, electroencephalographic records, sleep disorders, intellectual disability, recurrent infections, medical treatment options, response to treatment and follow-up time, and evaluations of psychosocial support were examined. Statistics were done with Turcosa and SPSS22 program.

Benefit from the therapy based on: Fomr antiepileptic drug: At least 50% reduction in the number, duration, and frequency of seizures. For Antipsychotics, ADHD treatment, and Melatonin: Benefit was accepted according to family or doctor's responses. No specific scale applied

RESULTS

In a total of 102 pediatric patients, the mean age at presentation was 51.2 months. 48.3% had seizures, 33.3% had development delay, typical phenotype and behavior was 6.9%, psychiatric symptoms easy excitability, sleep problems, and atypical autism) was 4.9%, isolated speech disorders were 2.9%, and attention-deficit admitted with hyperactivity disorder(ADHD) was 1 %. Patients not able to speak were 43%. Intellectual disability was 76.4%. Behavior problems were 86%; Extremely friendly, unusually happy behavior pattern %67.6, frequent and unusual laughter %55.7, clapping hands 47%, and difficulty walking was 90 %. Epilepsy frequency was 85%, and 61% was generalized. EEG rhythm was: Type1 persistent rhythmic 4–6Hz activity 32.8%, Type2 rhythmic delta 2–3Hz sharp spike-wave 45.2%, and Type 3 high amplitude 3-4 Hz sharp spike wave was 23 %.

CONCLUSİON

This study recommends that clonazepam/clobazam or valproate could be the first choice in antiepileptic therapy.

Tablo 1. Treatment Options For Angelman and Follow-up

Treatment Used		Number of Patients	*Benefit rate of the treatment method
Psychotherapy		44	%18
Special Education		71	
Antiepileptic Drugs	Totaly	88	
	Clonazepam/ Clobazam	**37**	**36/37 - 97.2%**
	Lamotrigine	**16**	**13/16 - 82.2%**
	Valproate	**68**	**53/68 - 77.9%**
	ACTH	**13**	**10/13 - 76%**
	Levetiracetam	43	22/43 - 53.6%
	Phenobarbital	23	10/23 - 43%
	Steroids	7	3/7 - 42%
Antipsychotics		26	9/26 - 34.6
ADHD treatment stimulant		19	10/19 - 52%
Melatonin		27	17/27 - 62.9
Follow up	Going on	83	81.3%
Mortality		1	0.9

*Benefit from the therapy based on:

For antiepileptic drugs: At least 50% reduction in the number, duration, and frequency of seizures

Antipsychotics, ADHD treatment, Melatonin: Accepted based on Family or Doctor's responses. No specific scale applied

REFERENCES

Clayton-Smith, Jill, and L. A. E. M. Laan. "Angelman syndrome: a review of the clinical and genetic aspects." Journal of medical genetics 40.2 (2003): 87-95.

Laan, Laura AEM, and Alla A. Vein. "Angelman syndrome: is there a characteristic EEG?." Brain and Development 27.2 (2005): 80-87.