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Autism Spectrum Disorder: One Year Review Cases

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Abstract

Autism Spectrum Disorder is a neurodevelopment disorder, with the etiology unknown in 75 - 80% of cases.

The aim of this review is find an etiology of ASD, detected in genetic consultation during a period of one year (1/6/2017 to 31/5/2018). We found 22 cases with this criteria according to DSM-V. A total of 9 cases (40.9%) of ASD was detected an etiology: 2 x fragile, 1 Angelman, 1 Rett, 1 Hunter, 1 trisomy 8, 1 del2p13 and 1 del 6p21.3. The gender ratio was 4.5 male/1 female. The average age of diagnosis was 4.7 years.

In patients with ASD, we need to try to find an etiology. Will be important for future support treatment, prenatal diagnosis and genetic counseling.

Keywords: *Autism Spectrum Disorder; Etiology*

Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopment disease.

The ASD according to the new definition by DSM-V, include autistic (autism), Asperger, childhood disintegrative disorder and pervasive developmental disturbance without further specification. The prevalence is 4(M)/1(F). In 75 - 80% of cases, its etiology is unknown [1].

Aim of the Study

The aim of this study is find an etiology of ASD, detected in genetic consultation during a period of one year

Material

Review all cases of ASD during a period of one year, from 1/6/2017 to 31/5/2018 in a genetic consultation of Centro Hospitalar Conde S Januário, Macau, China.

Methods

All cases with ASD, according to DSM-V, were screened for karyotype, X-fragile, microarray, metabolic disease (aminoacid, organic acid, lactic acid, pyruvic acid, ammonia, creatine and carbohydrate degradation transferrin-CDT). We also screened for Angelman syndrome and Rett disease in patients with absent speech or movement disorder.

We reviewed the age of diagnosis of ASD, gender, etiology of ASD and comorbidities of all cases detected.

Results

During this period we found 22 cases of ASD. The average age of diagnosis was 4.7 years. The gender ratio was 4.5 male/1 female. We found a total of 9 cases (40.9%) of ASD with etiology: 2 x fragile, 1 Angelman, 1 Rett, 1 Hunter, 1 trisomy 8, 1 del2p13 and 1 del 6p21.3.

The comorbidities associated with ASD were microcephaly, hypothyroidism, heart sick sinus syndrome and brain malformation (defect of corpus callosum and cerebellum) in 4 patients (Rett, 45X0, no etiology case and del 2p13, respectively) (Table 1).

Cases	Age of diagnosis of ASD (years)	Etiology	Comorbidities
1	7	No	No
2	7	No	No
3	5	No	No
4	2	Hunter	No
5	4	No	Sick sinus syndrome
6	2	No	No
7	4	No	No
8	3	No	No
9	15	Trisomy 8	No
10	13	No	No
11	3	Rett	Microcephaly
12	2	No	No
13	9	6p21.3	No
14	2	No	No
15	6	X fragile premutation	No
16	2	No	No
17	12	45X0	Hypothyroidism
18	8	No	No
19	2	X fragile premutation	No
20	3	No	No
21	3	Angelman	No
22	5	del 2p13	Defect of corpus callosum and cerebellum

Table 1: *Age of diagnosis, etiology and comorbidities of ASD cases.*

Discussion

Why is so important to find an etiology in ASD patients?

We can start a treatment (pharmacological and intervention strategies), clarify the prognosis situation, offer prenatal diagnosis and genetic counseling for the couple.

In our one year retrospective study, the number of cases detected in genetic consultation was more than one per month, almost two per month.

The gender ratio was approximately the same as in the literature.

The 9 cases detected with an etiology for ASD, we found 8 with chromosome disorders and only 1 with metabolic disease. The percentage of cases with etiology was 40.9%, more than we expected if we compared with the average prevalence (25 - 30%). Most of the metabolic cases are cause by respiratory chain disorder. In our case, we found a Hunter disease.

Depending on typical dysmorphic features, absent of speech and stereotyped movements disorders, we can do the molecular study of Angelman syndrome or Rett disease. In our one year data review, we found 2 cases [1-3].

Conclusions

In patients with ASD, at any age, we need to try to find an etiology. Will be important for future support treatment, prenatal diagnosis and genetic counseling.

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