Retrospective Evaluation of Cases Diagnosed with Classical Congenital Adrenal Hyperplasia Due to 21 Hydroxylase Deficiency

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ESPE 2021
The 59th Annual Meeting Online
22-26 September 2021

INTRODUCTİON

Congenital adrenal hyperplasia (CAH) is an autosomal recessive disorder. Diagnosis, monitoring of treatment, adjustment of drug doses are important for height, puberty and psychologic status of patients and their families.

AIM

Our aim was monitoring and reviewing of the cases in the follow-up of our clinic for 25 years which will provide important determinations and informations.

METHOD

- Patients with classical CAH due to 21-hydroxylase deficiency who were followed regularly in pediatric endocrinology clinic between 1994 and 2019 were included in this study. Informations of admission, follow-up, control and laboratory findings were obtained from electronic file data of patients.
- Cases diagnosed with classical 21-OH deficiency; were classified into two groups as simple virilizing type (SV) and salt wasting type (SW) according to the complaint at presentation, age at diagnosis, and clinical and laboratory findings.

RESULTS

- 48 cases
- 48% genotypically female, 30% of genotypically females' familes were told that their babies were male at birth
- 50% consanguinity+, 65% had similar family history of diesease, 38% had siblings with disease
- 75% SW, diagnosis at 55.8±76.8 day (5 day-10 months)
- 25% SV, diagnosis at 3.56±2.36 years (50 days-6 years 7 months)
- Vomiting (48%) and ambigius genitalia (44%, 93% of females) were the most common presenting complaints
- 42% had salt wasting, 58% had adrenal crisis at diagnosis
- 18% were diagnosed with standard dose ACTH stimulation test

Follow up

- 44% had good metabolic control
- 44% had corrective operation for ambigius genitalia (2 of 46XX cases were raised as male)
- 23% had treatment for puberty precocious
- Final height SD's were -2.57± 0.96 in SW group and -0.97±1.33 in SV group (p=0.029)
- (Hydrocortisone doses, BMI-SD values and target height SD values were similar)

Additional neuropsychological disorders
Epilepsy as a result of hypoglycemic convulsions (n=1)
Major depression (n=1)
Psychosis (schizophrenia) (n=1)
Anocrexia nervosa (n=1)
Autism (n=1)
Mild mental retardation (n=1)

Genetic	N(%)
I2G-IVS2-A/C homozygous	14 (%29)
Q318X homozygous	6 (%12,5)
Compound heterozygous	6 (%12,5)
Large deletion	4 (%8)
R356W homozygous	3 (%6)
Q318X heterozygous	3 (%6)
I172N homozygous	3 (%6)
P30L, I172N, V281L, Q318X, R356W homozygous	1 (%0,2)
V281L, Q318X, R356W homozygous	1 (%0,2)
Unknown	7 (%15)
87.5% genotype phenotype concordance (4 cases of P30L and I172N mutations with SW type and 2 cases of Q318X heterozygous and I2G homozygous with SV type)	

Additional disease due to CAH/treatment	Female (N)	Male (N)	SW (N)	SV (N)	Total (N)
Puberty precocious	6	5	8	3	11
Hypertension	5	3	6	2	8
Obesity	5	2	3	4	7
Short stature	3	3	4	2	6
Adrenal rest tumor	0	6	5	1	6
Enuresis	2	3	3	2	5
Insulin resistance	2	2	2	2	4

CONCLUSIONS

Close follow-up and psychological support is required for monitoring of chronic diseases like CAH.

CONTACT INFORMATION

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