The long term outcome of congenital adrenal hyperplasia due to 21-hydroxylase deficiency at KFSHRC-Retrospective study

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Background

- Congenital adrenal hyperplasia (CAH) refers to a group of autosomal recessive inherited disorders characterized by defects in cortisol synthesis.
- 21-hydroxylase deficiency is the most commonly defective enzyme representing more than 90% of cases. Affecting 1 of 8000 live births in Saudi Arabia.
- The deficiency of 21-hydroxylase enzyme results from mutations or deletions in the CYP21A2 gene found on chromosome 6p.
- 21-hydroxylase deficiency requires life-long steroid replacement therapy.
- Without appropriate monitoring, it may result in significant complications related to over or under replacement.

Aim of the Study

Assess the health status of adolescent & adults with 21-OH deficiency, the need for changes in current management of pediatric patients and to evaluate the need for endocrine specialist care as adult.

Methodology

It is a retrospective study. We reviewed medical files of all 21 hydroxylase deficiency cases still undergoing follow-up checks in pediatric clinics and are above the age of 14 years. All clinical, bio chemical, and genetic data were collected.

Results

Among the 79 patients involved in the study, 70% were females. Mean age for males is $16.8 \pm 4.6$ (range: 15-30 years), while mean age for females is $20.3 \pm 7.2$ (range: 15-41).

Molecular data of patients with 21 hydroxylase deficiency

- Petersen, C.-G.
- c.952 C>T:p.G318R

All patients with above mutation had classical salt wasting 21 hydroxylase deficiency

Current Medications

- Hydrocortisone
- Prednisone
- Dexamethasone

Prevalence of complications

UK-90 with 21-hydroxylase deficiency
- KFSHRC - 79 with 21-hydroxylase deficiency

- Median age 34 (range 18-49) years
- Median age 20 (range 15-41) years

Conclusion

- Control of androgens was highly variable with a high serum Androstenedione found in 40% of patients, whereas 31% had suppressed levels suggesting glucocorticoid over treatment.
- 22% were severely short (< -3 SD) (final height).
- 35% were obese.
- 11% had primary amenorrhea, 21% of male patients had Adrenal rest tumors.
- 5% had hypertension while hypercholesterolemia was present in 24%.
- Insulin resistance was found in nine patients out of 25 patients.
- Osteoporosis was present in 15% while osteopenia was present in 34%.

References