Rare Causes of Primary Adrenal Insufficiency at King Faisal Specialist Hospital - Retrospective Study

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Introduction:
- Adrenal insufficiency is a rare, but potentially fatal medical condition.
- In children, the cause is most commonly congenital adrenal hyperplasia (CAH) but in recent years a growing number of causative gene mutations have been identified resulting in syndromes that share primary adrenal insufficiency (PAI) characteristics.
- PAI incidence in Europe reported 82–144/million.
- Higher CAH incidence has been reported in Saudi Arabia (1/7908) and 44 cases have reported for other causes.
- Underlines causes for a lot of cases have not been identified till Whole exome sequencing have been applied.

Objectives:
- Primary: To identify causes for primary adrenal insufficiency at KFSHRC.
- Secondary: To identify causative genes and common features of PAI.

Methodology:
- Study design is retrospective cross-sectional study by reviewing medical records.
- Inclusion criteria: All patients following with Pediatric Endocrinology clinics at KFSHRC during 2018 with PAI.
- Exclusion criteria: All cases of congenital adrenal hyperplasia and autoimmune polyendocrine disease were excluded.
- IRB approved the research with RAC Number: (2181257).
- Data were collected and entered using Excel Sheet then analyzed by SPSS.

Result and Discussion:
- The most common causes of PAI are Adrenoleukodystrophy then Familial Glucocorticoid Deficiency and Adrenal Hypoplasia which is different from Hsieh and White study.
- X-linked diseases account for 56% of them which explains predominance of male on the study.
- Adrenoleukodystrophy is common on our hospital due to availability of transplant and screening of other family members is recommended.
- ACTH resistance and Adrenal Hypoplasia present early on life but majority present late.

Conclusion:
- Causes other than CAH should be suspected during evaluation of primary adrenal insufficiency.
- Whole exome sequencing helped in diagnosing majority of cases.
- More researches are needed to identify common genes in our society for developing Primary Adrenal Insufficiency panel.
- All male patients presenting with PAI to be screened for Adrenoleukodystrophy (VLCCFA) and Adrenal Hypoplasia (CK, Lipid, US, DAX1 gene). Also, Karyotyping is recommended for female patients.
- Consider screening all patients with PAI for proteinuria to rule out Nephrotic Syndrome type 14.

References: