Prevalence Of 21-OH CAH: In our cohort of CAH (186 patients from 165 families), 21-OHD CAH was diagnosed and confirmed by clinical and biological grounds in 168 patients from 145 families, rendering it the most common of CAH before 3 beta-hydroxysteroid-dehydrogenase deficiency (12 patients from 8 families) and 11-hydroxylase deficiency (6 families, 8 patients).

Of 168 patients, 114 were Females (F) and only 54 were Males (M), Sex Ratio was 1M/2F. Consanguinity was found in 61% of the cases. The classical salt-wasting form (SW) of 21-OHD CAH was most common (80%) comprising 135 (66%) patients, while 31 patients (77%) had simple virilising (SV-21OHD CAH) and 2 Females had non-classical (NC-21OHD CAH) of CAH (being untreated) as seen in Fig 1 and 2.

Initial referral (cf Fig 3) was with disorder of sex development (SDS) in 27% (all females), dehydration in 28% (6 males); the association of SDS and Dehydration in 35% (100% Females); sexual precocity (7%) and family history of CAH. At presentation, SDS was found in all but 46.4KX patients. 2 NC-21OHD (2 treated prematurely). Severe hyponatremia and dehydration occurred in 69 cases (31.5% neotekat males and females (61 vs 45%, p=0.06).

Ages at presentation and at the initiation of hydrocortisone (Table 1) were significantly later in males than females (191±524 vs 83±331 days and 268±639 vs 153±152 days, p=0.001), attributable to SDS being the presenting feature in the latter. Mean age at diagnosis was significantly earlier in the SW (51±136 days) than in the SV-21OHD (617.9±397 days). Fludrocortisone (being unavailable in Algeria) was started in 148 patients at 140±308 days (vs 118±406 days for hydrocortisone in all patients).

Mean plasma Sodium at diagnosis (Table 2) was lower in males than females (118±10 vs 123±10 meq/l, p=0.03) and between the SW and the SV-21OHD (120±10 vs 130±3 meq/l, p=0.01).

Hormonal data (Table 2): 17OH-Progesterone (17OHP) was elevated in all patients (1672±3048 nmol/l), there was no significant difference between both sexes and between the 2 classical forms. Renin was significantly higher in the SW 21-OHD (2817±6483 vs 153.2±165 mg/l). p=0.001.

Sex assignment: 62 of 114 (54.4%) 4KX patients were initially assigned as males, mean±SD (range) at reassigment was 3.8±0.36 months in 58 patients with 4 46XX patients raised as males according to parental wishes.

Treatment: All patients were treated with hydrocortisone, 148 (88%) were initially treated with Fludrocortisone. Only 30.8% of the females had undergone surgery at a mean±SD (range) age of 3.6±4.5 (0.5-10) years.

Clinical outcome (Table 3): Since diagnosis, 8 (4.7%) patients have died in adrenal crisis. Mean age at last visit was 5.3±4 years. More than one third (37.5%) of the Patients have presented at least one episode of dehydration since diagnosis, 24% have advanced bone age, 14% have been treated with LHRH agonists and 13% with growth hormone, 8 males have testicular calcifications and 3 patients have adrenal calcifications. Finally, of 89 patients aged >4 years, 18 (20%) have moderate to severe mental delay.

CONCLUSIONS

Currently, males with 21-OHD are diagnosed half often as females, reflecting death from SW during the first few weeks. Delay age at diagnosis causes severe hypoglycaemia in SW patients, which increase the risk of mortality and developmental delay. National screening for CAH in Algeria and all Maghreb countries where consanguinity rate and hence CAH prevalence high is urgently required.

To examine age at presentation and outcome in children diagnosed with 21-hydroxylase deficient congenital adrenal Hyperplasia (21-OHD CAH) in Algeria in the absence of a national newborn screening program.

This was a retrospective analysis of patients followed in a single centre from 2007 to 2017. The diagnosis of CAH was established on clinical and biochemical grounds ± genetic analysis. Statistical analysis was done with Epi-info 7 (CDC) software.