CONGENITAL ADRENAL HYPERPLASIAS: NEONATAL REVELATION

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Introduction:

- The term congenital adrenal hyperplasia (CAH) encompasses a group of autosomal recessive disorders, each of which involves a deficiency of an enzyme involved in the synthesis of cortisol.
- Includes several clinical forms.
- Short-term prognosis good: if diagnosis and early management.

Materials & Methods :

•Retrospective study, carried out in Neonatal Intensive Care Unit, Mohammed VI University Hospital; MARRAKESH

•During a period from July 2015 - April 2017.

•The population of our study included all neonates diagnosed with congenital adrenal hyperplasia.

Results:

•We collected nine cases, which were included during the study period.

•Mean age at diagnosis: 13.4 days.

•The sex ratio was 0.6 (fig.1) .

•The marriage was consanguineous in three cases.

•Vomiting associated with diarrhea: the most common signs.

•The 17-OHP: 07 cases did the 17-OHP assay and showed a high level of the 17-OHP

•HCS diagnosis by 21 hydroxylase deficiency was retained, the cortisol assay was normal in both cases which did the dosage.

•The karyotype was performed in 8 cases

•Abdominal-pelvic ultrasound: Concurrent with karyotype data.

•Treatment: All our patients had benefited from rehydration, correction of electrolyte disorders and intravenous hydrocortisone hemisuccinate hormone therapy, combined with long-term treatment with:

- Hydrocortisone: 20 mg / m² / day in 2 to 3 doses taken orally, relying on the intravenous route.
- Fludrocortisone: 40 to 100 µg / day in the classic form with loss of salt.
- NaCl supplementation at a dose of 500mg x 3 / d orally until the age of 2 years.

Figure I: Distribution by sex of patients

Study	Number of cases with 21-OH deficiency	Consanguinity	Age at the time of diagnosis	Sex ratio (boy/girl)
Univerity Hospital IBN ROCHD (Casablanca)	8/8	1/2	22 days	1
Univerity Hospital HASSAN II (Fez)	6/9	1/2	43 days	5
Our study (Marrakesh)	7/9	1/3	13 days	0.6

Table I: Comparison of the epidemiological data between the study conducted at the Ibn Rochd University Hospital in 2002, the study carried out at Hassan II University Hospital Fes 2011 and our study.

Discussion:

• CAH is defined by a deficiency of one of the enzymes, causing according to the enzyme reached, a lack of synthesis of one or more hormones and sometimes an excess of synthesis of other hormones.

•Deficiency of 21-hydroxylase: 90 to 95% of HCS, prevalence: 1/15000 birth with variation according to ethnic groups.

•11β-hydroxylase deficiency: 5 to 8% of HCS

•In Morocco, the prevalence of congenital adrenal hyperplasia is not yet known, given the lack of a national screening program

• In 21-hydroxylase deficiency, there are classic clinical forms (CFs) that are severe and non-classical clinical forms (NCFs) of moderate severity in 21-OH deficiency.

• The incidence of CF is 1 / births including 67% of forms with salt loss and 33% of pure virilizing forms without salt loss.

• Emergency treatment consists of the correction of dehydration or shock, associated with substitution and brake treatment.

•Therapeutic education :

•In case of stress or intercurrent illnesses: The doses of Hydrocortisone must be doubled or tripled

•Monitoring is required through regular checkups (D15, D45 then every 3 months for 2 years then every 6 months), with clinical-biological evaluation of efficacy and tolerability of treatment

Conclusion:

•Screening at birth for congenital adrenal hyperplasia due to a 21-hydroxylase deficiency should be done systematically, because in the absence of an abnormality of EMB the only revelation is a salt loss syndrome.

•Hormone replacement and lifelong therapy requires rigorous monitoring that can improve prognosis, avoid complications related to chronic hyperandrogenism and ensure normal fertility.

•Genetic counseling in its place.

•Antenatal screening: not yet done in our context

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