

# CONGENITAL ADRENAL HYPERPLASIA IN DETAIL: A MULTICENTRIC AND INTERDISCIPLINARY STUDY IN MINORS WITH DIFFERENT SEXUAL DEVELOPMENT SEXUAL



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## INTRODUCTION

Among the entities that involve a different sexual development (DSD), cases of congenital adrenal hyperplasia (CAH) are the most frequent. Even so, there are still controversial points in its clinical care today..

## AIM

We present the HSC in detail after the general study of all the DSD in pediatric age.

## METHOD

Descriptive multicenter study by retrospective review (2006-2019).

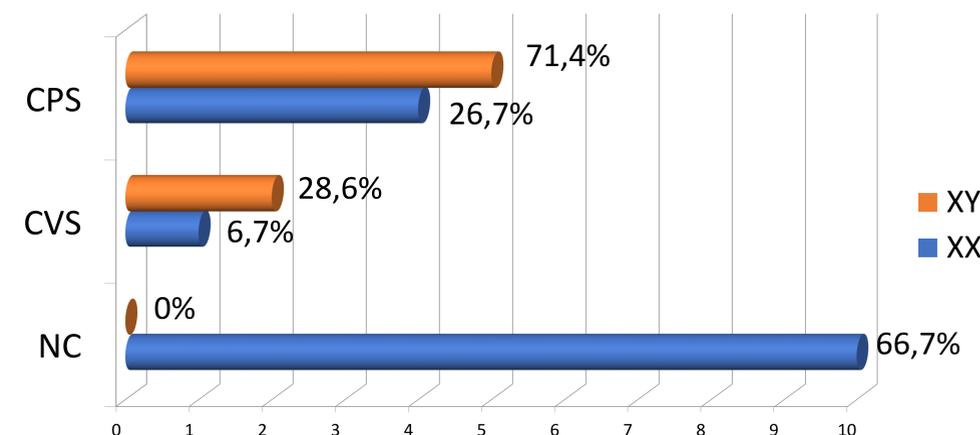
Statistics performed with non-parametric tests due to the small sample size.

Interdisciplinary assessment through satisfaction surveys to families / patients.

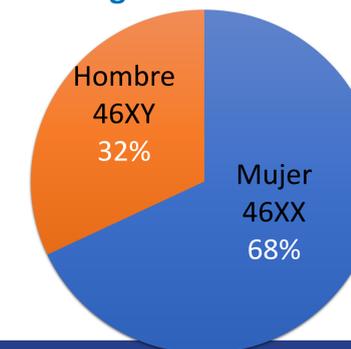
## RESULTS

Of the 67 included DSD patients, 33% were HSCs. 68% women and 32% men, with a total concordance between legal sex, current gender and chromosomal sex. Median age: 10.5 years, IQR 8-14. The most frequent clinical form was non-classical (NC) 45.5%, followed by classical with saline loss (CPS) 41%. The distribution of each clinical form according to the karyotype is significant ( $p = 0.013$ ), the most frequent with XY karyotype is CPS and in NC forms all cases are XX. Median age at the start of the study: 2.5 months, IQR 0.75-42.5. In 68% the diagnostic process began before 12 months of age. According to gender, when comparing the medians, the results are not significant ( $p = 0.187$ ). On the other hand, there is a higher median age in men with simple virilizing clinical forms (CVS), with 31.5 months. All cases were genetically defined, median age 1 month, IQR 0-5.2; a de novo mutation (I172N) was even identified. The clinical form with the earliest diagnosis was CPS, IQR 0-1.5 months. Detailing the associated pathologies, the following stand out: 12 uro-genital sinuses, 2 vesico-ureteral refluxes, no cases of testicular maldescent or TARTs, 3 ovarian bands, 3 cases of precocious pubarche, one case of acne and 3 hydroelectrolytic decompensations. No metabolic disorders or osteopenia were described. Regarding the treatment, in no case was prenatal treatment offered. 86.3% are under medical treatment, 23% with glucocorticoids in monotherapy. The NC forms are those with the greatest variability with 30% without treatment, 50% glucocorticoids and 20% mineralo-glucocorticoids, significant differences ( $p = 0.005$ ). Three cases required psycho-psychiatric treatment, one CPS and two NC. It should be noted that the only two patients who required hospital admission and pharmacological treatment of all DSD were two women with CAH, without a diagnosis of gender dysphoria. 16 surgical procedures were performed, 68% for diagnostic purposes and 7 genitoplasties. Median age of genitoplasties: 29 months, IQR 20.5-37. Registering 6 complications. Regarding the surgical result, 1/3 of patients / family declared it excellent and 1/3 acceptable. While 56% were good-excellent according to surgeon / a. Assessing the specialist teams, all classify endocrinology and surgical teams as good-excellent, as opposed to psychological assistance (55% bad-terrible and 45% good-excellent).

Cariotype and type of HSC



Actual gender & chromosomal



## CONCLUSIONS

In all cases, the responsible mutation has been identified, even a de novo one. Unlike the literature, the most common clinical form is CN. Only three cases of adrenal decompensation were described, without prenatal screening, thanks to early diagnosis. The post-surgical assessment is consistent according to the surgeon and family / patient. Major surgical procedures are scarce, the majority being diagnostic procedures.

**We advocate for psychological assistance as the main point of improvement**

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