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P3-P028

# DISCRETE VIRILIZATION IN GIRLS WITH THE CLASSIC FORM OF CAH: THE IMPORTANCE OF A DETAILED GENITAL EXAMINATION AT BIRTH

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## Introduction

**FOETAL SERUM ANDROGEN CONCENTRATION ➔ EXTERNAL GENITALIA**

**CONGENITAL ADRENAL HYPERPLASIA CLASSIC FORMS (21OHD) ➔ FEMALE GENITAL AMBIGUITY MOST FREQUENT CAUSE**

**CLASSIC FORMS VIRILIZATION: SALT LOSER (SL) OR SIMPLE VIRILIZING (SV) & NONCLASSIC FORMS (NC)**

**VIRILIZATION (PHENOTYPE) ≈ ENZYMATIC DEFICIENCY**

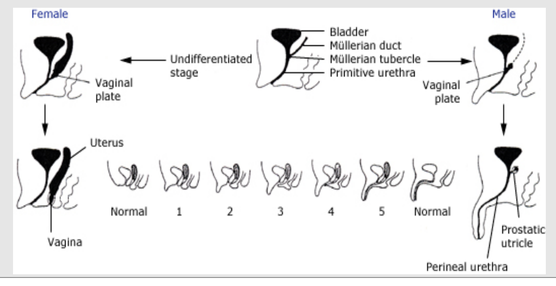
- **OBJECTIVE: TO DESCRIBE 21OHD FEMALES WITH LATE DIAGNOSIS DUE TO DISCRETE VIRILIZATION**

## Methods

**RETROSPECTIVE DATA: AGE AT DIAGNOSIS, 17OHPROGESTENONE, BONE AGE**

**GENITAL AMBIGUITY ➔ PRADER CRITERIA**

**MOLECULAR STUDY ➔ PREDICTED PHENOTYPE**




## Cases report

**Case 1 => first consultation 44m: precocious pubarche (2y) and clitoromegaly (without labial fusion)**

**Case 2 => first consultation 31m; case 1 sister's; precocious pubarche (1y) and clitoromegaly (without labial fusion)**

**Case 3 => first consultation 7m; CAH NSL sister's; labial fusion since birth**

**Case 4 => first consultation 19m; precocious pubarche (3m), clitoromegaly and labial fusion since birth**

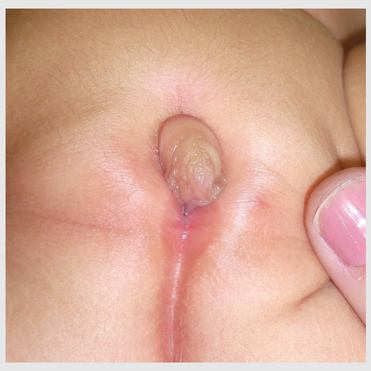
**Case 5 => first consultation 23m; clitoromegaly and labial fusion since birth (surgically corrected)**

**Case 6 => first consultation 27m; clitoromegaly since birth (without labial fusion)**

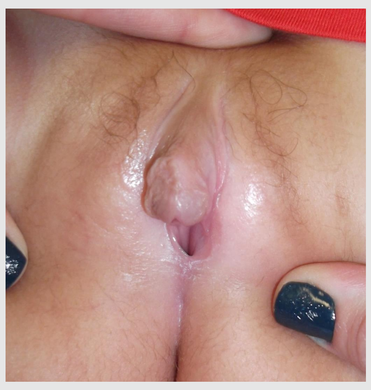
**The molecular study showed that all children carried a salt-wasting and a simple virilising mutation in either allele (Allele-SW and Allele-SV), being, therefore, the predicted phenotype SV.**

**Age at diagnosis (months) - Age, Bone age / Chronological age (years) – BA/CA, Height (z-sc), genital virilisation (Prader criteria) - Prader, 17OHP serum levels (ng/mL) - 17OHP, mutations (Allele-SW and Allele-SV)**

| | Age | Height | Prader | 17OHP | BA/CA | Phenotype | Allele SW | AlleleSV |
|---|---|---|---|---|---|---|---|---|
| 1 | 44 | - 0.05 | 2 | 79.5 | 5.2/3.8 | SV | c.-315C>T;c.290-13A/C>G | c.-103A>G;p.Ile172Asn |
| 2 | 31 | 0,09 | 1 | 158.8 | 5.6/2.9 | SV | c.-315C>T;c.290-13A/C>G | c.-103A>G;p.Ile172Asn |
| 3 | 7 | - 0.45 | 3 | 82.0 | - | SV | Large gene conversion | p.Ile172Asn |
| 4 | 19 | 1.15 | 3 | 224.4 | 3.8/1.9 | SV | p.Arg356Trp | p.Ile172Asn |
| 5 | 23 | 2.65 | 1* | 206.8 | 5.1/2.4 | SV | p.[Leu308Phefs*6;Gln318*;Arg356Trp] | p.Ile172Asn |
| 6 | 27 | 0.79 | 1 | > 50.0 | 5.3/2.2 | SV | p.[Gln318*; Arg356Trp] | p.Ile172Asn |



Case 3



Case 4

## Coments

- **We report six cases of CAH due to 21OHD, all with the classic, simple virilising form confirmed by molecular study, whose discrete genital virilisation was not identified at birth by the paediatrician.**
- **According to the relatives, all of them had clitoromegaly and in 3 cases there was also labial fusion at birth.**
- **Through this presentation, we would like to draw attention to the importance of a careful examination of the external genitalia at birth, in order to identify small alterations and obtain early 21OHD diagnosis.**