steroid 17-20 desmolase deficiency

Eponyms: pseudohermaphroditism male
steroid 17,20 desmolase deficiency

Inheritance: supposed autosomal recessive

Semeiological Synthesis: Endocrino-genital disorder. Ambiguous genitalia in 46,XY male, without adrenogenital syndrome; low excretion of all androgens, and normal glucocorticoids/mineralocorticoids secretion; infertility/delayed puberal development in female, because of estrogen deficiency.

Group
Sub group
Signs:

GENITAL DISORDERS
- genital dysfunctions
  - infertility, sterility
  - genitalia, ambiguity
  - ambiguous genitalia, male
  - pseudohermaphroditisms, male

LABORATORY DATA
- biochemical markers
  - metabolic defect
- plasma proteins, anomalies
  - hormones, dysfunctions

Super group:

Super aggrec. Aggregations:

METABOLIC DISORDERS
- hormones, dysfunctions

PSEUDO-HERMAPHRODITISM
- pseudohermaphroditism, male

Differential diagnosis:
- 741 adrenal hyperplasia, Peterson type
- 28874 precocious puberty, male limited
- 21605 progesterone resistance
- 21785 pseudohermaphroditism, male, LH molecule defect

Bibliography
OMIM ID: 309150