Title: Reversal of primary male infertility and testicular adrenal rest tumors in salt-wasting congenital adrenal hyperplasia.

Authors: Collet TH, Pralong FP

Journal: The Journal of clinical endocrinology and metabolism

Year: 2010 May

Issue: 95

Volume: 5

Pages: 2013-4

DOI: 10.1210/jc.2009-2691

IMAGE IN ENDOCRINOLOGY

Reversal of primary male infertility and testicular adrenal rest tumors in salt-wasting congenital adrenal hyperplasia

Tinh-Hai Collet[1], François P Pralong[2]

Services of Internal Medicine[1] and Endocrinology, Diabetology and Metabolism[2], Department of Internal Medicine, University Hospital, Lausanne, Switzerland

Corresponding author

François P Pralong, MD
Service of Endocrinology, Diabetology and Metabolism
University Hospital
Bugnon 46
1011 Lausanne
Switzerland
Phone: +41 21 314 0596
Fax: +41 21 314 0597
Email: francois.pralong@chuv.ch

Short title

Testicular adrenal rest tumors and primary infertility in a male patient with a salt-wasting form of congenital adrenal hyperplasia are reversible with one year of corticoid replacement therapy.

Keywords

Fertility, male, congenital adrenal hyperplasia, testicular adrenal rest tumor

Word count

260 words in the text (limit 250)
50 words in the caption (limit 50)

Declaration of interest

The authors declare that there is no conflict of interest that could be perceived as prejudicing the impartiality of the research reported.

Funding

This research did not receive any specific grant from any funding agency in the public, commercial or not-for-profit sector.

Text

A 26-year old male with **a** salt-wasting form of congenital adrenal hyperplasia **(CAH)** presented with **a 4-year history of** primary infertility. **He had done** well on **deflazacort (range 6-18mg per day) and fludrocortisone (0.05mg bid)** until late adolescence when he had several Addison's crises due to decreased therapeutic **adherence**. **He was lost to follow-up for several years prior to infertility work-up.** LH and FSH **were undetectable while serum testosterone level was** 37.4nmol/L (N: 8-26), ACTH 501ng/L (N: 10-60) and 17-OH progesterone 1047nmol/L (N: 1.8-9.2). CT-scan showed massive bilateral adrenal hyperplasia (Figure), and testicular ultrasonography multiple bilateral testicular adrenal rest tumors (TARTs). Semen analysis showed azoospermia. **CAH was shown to be due to a missense mutation in** exon 8 of the 21-hydroxylase gene.

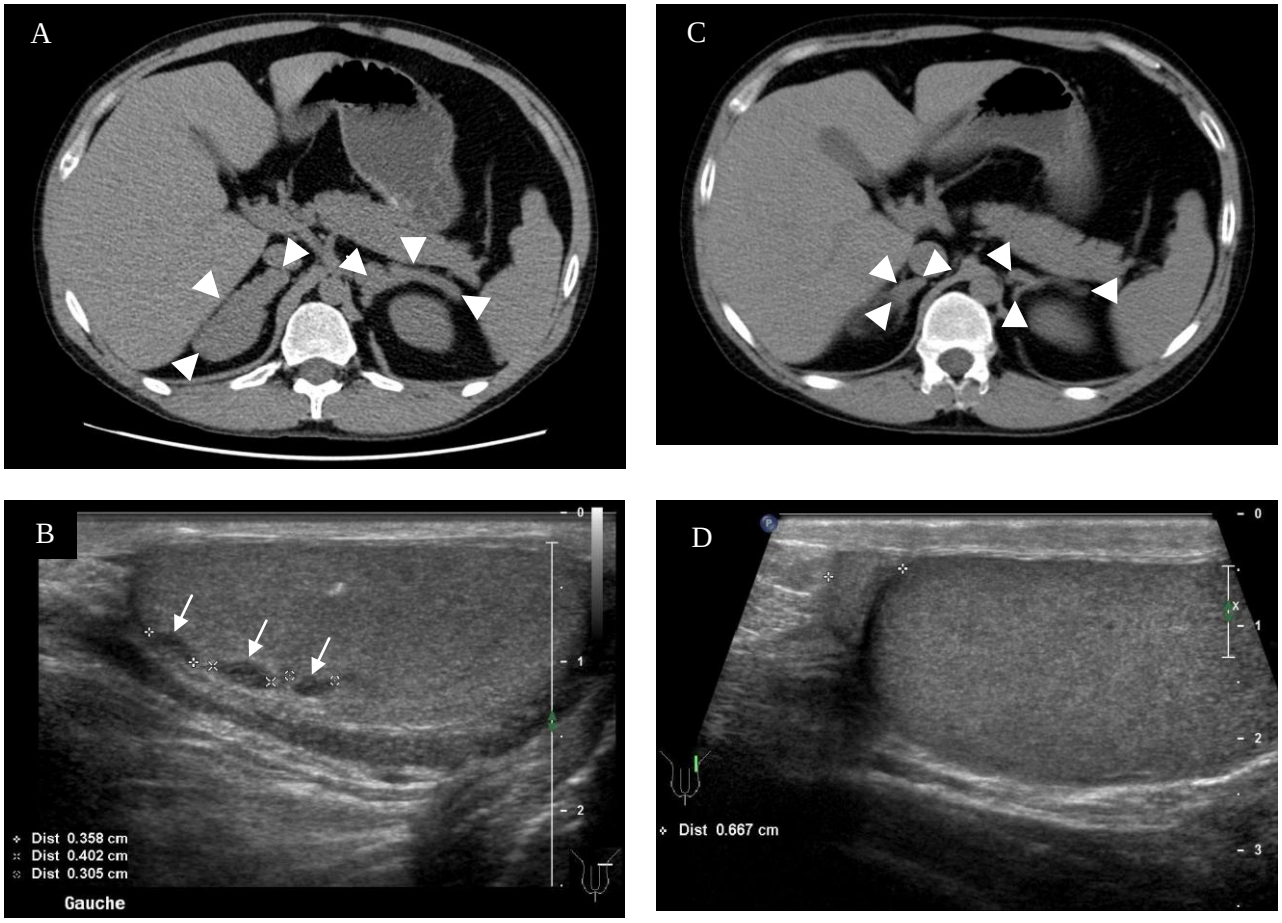
In response to dexamethasone (0.5mg bid) and fludrocortisone (0.05mg bid), ACTH and 17-OH progesterone levels **fell (8ng/L and 2.5nmol/l, respectively) while** LH (4.2U/L) and FSH (9.3U/L) **rose, followed by spontaneous conception. The patient's wife delivered a healthy term male baby 12 months after the change of replacement therapy. However, repeat semen analysis and paternity confirmation were not performed.** Repeat imaging studies after **13** months **of** treatment disclosed dramatic reduction of adrenal hyperplasia and the disappearance of TARTs.

These data demonstrate the rapid reversibility of TARTs using dexamethasone as **first** described by Cunnah (1), **without** side effects in contrast to previous reports (2). The chronological association of TART disappearance with pregnancy suggests **that mechanical obstruction on seminal tubules might have caused** this patient's infertility, **although gonadal axis suppression may also have contributed to infertility, as discussed recently (3, 4).**

Acknowledgments

The authors wish to thank Dr Fulgencio Gomez, MD, for previous care of the patient and helpful suggestions for the preparation of this manuscript.

Figure



After 4 years of infertility, CT-scan showed adrenal hyperplasia (**arrowheads**, A), and testicular ultrasonography multiple bilateral **3-4mm long** testicular adrenal rest tumors (TARTs, **arrows**, B). The introduction of dexamethasone and fludrocortisone and increased therapeutic **adherence** led to reduction of adrenal **size** (**arrowheads**, C) and TART disappearance (D) **after 13 months**.

References

1. **Cunnah D, Perry L, Dacie JA, Grant DB, Lowe DG, Savage MO, Besser GM** 1989 Bilateral testicular tumours in congenital adrenal hyperplasia: a continuing diagnostic and therapeutic dilemma. Clin Endocrinol (Oxf) 30:141-147
2. **Claahsen-van der Grinten HL, Otten BJ, Sweep FC, Hermus AR** 2007 Repeated successful induction of fertility after replacing hydrocortisone with dexamethasone in a patient with congenital adrenal hyperplasia and testicular adrenal rest tumors. Fertil Steril 88:705 e705-708
3. **Reisch N, Flade L, Scherr M, Rottenkolber M, Pedrosa Gil F, Bidlingmaier M, Wolff H, Schwarz HP, Quinkler M, Beuschlein F, Reincke M** 2009 High prevalence of reduced fecundity in men with congenital adrenal hyperplasia. J Clin Endocrinol Metab 94:1665-1670
4. **Claahsen-van der Grinten HL, Otten BJ, Hermus AR, Sweep FC, Hulsbergen-van de Kaa CA** 2008 Testicular adrenal rest tumors in patients with congenital adrenal hyperplasia can cause severe testicular damage. Fertil Steril 89:597-601