Journal of Andrology, Vol. 24, No. 4, July/August 2003

Y Chromosome Deletions in Azoospermic Men in India

KUMARASAMY THANGARAJ*, NALINI J. GUPTA†, KADUPU PAVANI*, ALLA G. REDDY*, SUBBAYA SUBRAMAINAN*, DEEPA SELVI RANI*, BIBASWAN GHOSH†, BAIDYANATH CHAKRAVARTY† AND LALJI SINGH*

*From the * Centre for Cellular and Molecular Biology, Uppal Road, Hyderabad, India; and the† Institute of Reproductive Medicine, DD 18/5/1 Sector 1, Salt Lake, Kolkata, India.*

Correspondence to: Dr Lalji Singh, Centre for Cellular and Molecular Biology, Uppal Road, Hyderabad, 500 007, India (e-mail: lalji{at}ccmb.res.in).

Genetic factors cause about 10% of male infertility. Azoospermia factors (AZFa, AZFb, AZFc) are considered to be the most important for spermatogenesis. We therefore made an attempt to evaluate the genetic cause of azoospermia, Y chromosome deletion in particular, in Indian men. We have analyzed a total of 570 men, including 340 azoospermic men and 230 normal control subjects. DNA samples were initially screened with 30 sequence-tagged site (STS) markers representing AZF regions (AZFa, AZFb, AZFc). Samples, with deletion in the above regions were mapped by STS walking. Further, the deletions were confirmed by Southern hybridization using the probes from both euchromatic and heterochromatic regions. Of the total 340 azoospermic men analyzed, 29 individuals (8.5%) showed Y chromosome deletion, of which deletion in AZFc region was the most common (82.8%) followed by AZFb (55.2%) and AZFa (24.1%). Microdeletions were observed in AZFa, whereas macrodeletions were observed in AZFb and AZFc regions. Deletion of heterochromatic and azoospermic regions was detected in 20.7% of the azoospermic men. In 7 azoospermic men, deletion was found in more than 8.0 Mb spanning AZFb and AZFc regions. Sequence analysis at the break points on the Y chromosome revealed the presence of L1, ERV, and other retroviral repeat elements. We also identified a ~240-kb region consisting of 125 bp tandem repeats predominantly comprised of ERV elements in the AZFb region. Histological study of the testicular tissue of the azoospermic men, who showed Y chromosome deletion, revealed complete absence of germ cells and presence of only Sertoli cells.

Key words: Male infertility, STS markers, repeat sequence

This article has been cited by other articles:

Journal of ANDROLOGY HOME

V. Suryavathi, A. Khattri, K. Gopal, D. S. Rani, S. Panneerdoss, N. J. Gupta, B. Chakravarty, M. Deenadayal, L. Singh, and K. Thangaraj
Novel Variants in UBE2B Gene and Idiopathic Male Infertility
J Androl, September 1, 2008; 29(5): 564 - 571.
[Abstract] [Full Text] [PDF]

Molecular Human Reproduction HOME

C.-H. Tsai-Morris, E. Koh, Y. Sheng, Y. Maeda, R. Gutti, M. Namiki, and M. L. Dufau
Polymorphism of the GRTH/DDX25 gene in normal and infertile Japanese men: a missense mutation associated with loss of GRTH phosphorylation
Mol. Hum. Reprod., December 1, 2007; 13(12): 887 - 892.
[Abstract] [Full Text] [PDF]

Journal of ANDROLOGY HOME

L. Hadjkacem-Loukil, I. Ayadi, A. Bahloul, H. Ayadi, and L. Ammar-Keskes
Tag STS in the AZF Region Associated With Azoospermia in a Tunisian Population
J Androl, September 1, 2007; 28(5): 652 - 658.
[Abstract] [Full Text] [PDF]

Journal of ANDROLOGY HOME

S. Rajender, V. Rajani, N. J. Gupta, B. Chakravarty, L. Singh, and K. Thangaraj
No Association of Androgen Receptor GGN Repeat Length Polymorphism With Infertility in Indian Men
J Androl, November 1, 2006; 27(6): 785 - 789.
[Abstract] [Full Text] [PDF]

Molecular Human Reproduction HOME

S. Rajender, V. Rajani, N. J. Gupta, B. Chakravarty, L. Singh, and K. Thangaraj
SRY-negative 46,XX male with normal genitals, complete masculinization and infertility
Mol. Hum. Reprod., May 1, 2006; 12(5): 341 - 346.
[Abstract] [Full Text] [PDF]

Journal of ANDROLOGY HOME

R. Singh, S. R. Deepa, S. Madhavi, N. J. Gupta, B. Chakravarty, L. Singh, and K. Thangaraj
Male Infertility: No Evidence of Involvement of Androgen Receptor Gene Among Indian Men
J Androl, January 1, 2006; 27(1): 102 - 105.
[Abstract] [Full Text] [PDF]