GMR Genetics and Molecular Research
Online Journal
ISSN:1676-5680

Androgen receptor gene CAG repeat polymorphism and risk of isolated hypospadias: results from a meta-analysis

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Genet. Mol. Res. 14 (1): 1580-1588 (2015)
Received January 10, 2014
Accepted March 12, 2014
Published March 6, 2015
DOI http://dx.doi.org/10.4238/2015.March.6.5

ABSTRACT. Studies investigating the association between the CAG repeat polymorphism and the risk of isolated hypospadias have reported conflicting results. The aim of this study was to quantitatively summarize the evidence for such a relationship. Two investigators independently searched the Medline, Embase, CNKI, and Wanfang databases. Weighted mean difference and 95% confidence intervals for the CAG repeat polymorphism and isolated hypospadias were calculated using a random-effects model. Subgroup analyses were performed by race, study design, sample for DNA extraction, and hypospadias classifications. This meta-analysis included 6 case-control studies, including 444 isolated hypospadias cases and 727 controls. The results showed that patients with isolated hypospadias had longer CAG repeats in their androgen receptor gene sequence (weighted mean difference = 1.36, 95% confidence interval = 0.60-2.13; P = 0.0005). Similarly, stratified analyses also detected significant associations in all subgroups, excluding the group with severe hypospadias (weighted mean difference = 0.35, 95% confidence interval = -0.42-1.12; P = 0.38). This meta-analysis indicated that longer CAG repeats were

associated with the risk of isolated hypospadias, and that longer CAG polymorphisms may be related to the etiology of isolated hypospadias. Future studies based on Asian and African-American patients should be performed to re-evaluate this association.

Key words: *AR* gene; CAG repeat polymorphism; Isolated hypospadias; Meta-analysis

