

Association of a single nucleotide polymorphism of the *DAZL* gene with susceptibility to spermatogenic failure

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INTRODUCTION: To investigate the role of *DAZL* in spermatogenic failure

METHODS:

DESIGN: case-control study and molecular analysis

SETTING: Medical college laboratory

SUBJECTS: 160 infertile Taiwanese men presenting with severe oligozoospermia and non-obstructive azoospermia and 116 fertile men

INTERVENTION: none

MAIN OUTCOME MEASURES: Testicular phenotypes of infertile men

RESULTS: An mutation located within the RNA-binding motif of the *DAZL* protein (386 A→G) in exon 3 was identified. It will lead to Thr54→Ala change (T54A) of *DAZL* protein. The frequencies of T54A allele in patients and control group were 7.39% and 0.86%, respectively ($p = 0.0003$). The phenotypes varied significantly in cases with heterozygous T54A polymorphism, ranging from hypospermatogenesis and maturation arrest to Sertoli cell-only syndrome. A combination of *DAZ* gene deletion and T54A polymorphism did not worsen the phenotype.

CONCLUSIONS: Our findings provide strong evidence for the role of the autosomal *DAZL* gene in human spermatogenesis. Heterogeneity of phenotypes in patients with T54A variant indicates involvement of complex genetic components in spermatogenic failure.