Objective: The aim of this study is to determine the prevalence of Y chromosome microdeletions in infertile Chilean men.

Material and methods: A group of 102 infertile men with azoospermia or severe oligozoospermia were screened while attending a fertility clinic for microdeletions in the azoospermia factor (AZF) region of Y chromosome by multiplex polymerase chain reaction. Genomic DNA was extracted from peripheral blood samples. Each patient was analysed for the presence of sequence tagged sites in the AZFa, AZFb, and AZFc regions.

Results: Azoospermia and severe oligozoospermia was found in 67 and 35 patients, respectively. Microdeletions were found in 9.8% of patients. The most prevalent mutation was AZFc, affecting 3.9% of the sample. This was followed by AZFbc with 2.9%, AZFa with 2.0%, and AZFb with 1.0%. Only azoospermic men were found to have these genetic alterations.

Conclusions: Prevalence of Y chromosome microdeletions in infertile Chilean men is similar to the prevalence presented in international studies. As AZFa and AZFb mutations are associated with complete absence of viable gametes, and AZFc has important consequences in the fertility potential of the offspring, these mutations have to be searched when presented with an infertile patient with severe sperm alterations. (C) 2015 Asociacion Espanola de Andrologia, Medicina Sexual y Reproductiva. Published by Elsevier Espana, S.L.U. All rights reserved.