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Sex Determination in Children with Ambiguous Genitalia by Polymerase Chain Reaction (PCR)

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ABSTRACT Sex determination in infants and children with ambiguous genitalia usually necessitates time-consuming and costly karyotyping. We have evaluated a simple, rapid and reliable method of postnatal sex determination by amplification of X and Y specific microsatellite markers DXS6797 and SRY respectively by polymerase chain reaction (PCR). Three probands M78, M59 and M61 with ambiguous genitalia were investigated. M78 showed a female complement of 46,XX while M59 and M61 have male complement of 46,XY. M78 was diagnosed as female pseudohermaphrodite while M59 and M61 were identified as male pseudohermaphrodite. Except M78, where a SRY sequence was identified by polymerase chain reaction (PCR), the results of PCR were in agreement with those of cytogenetic analysis. The present study reaffirms the fact that the polymerase chain reaction (PCR) based sex determination is more sensitive, rapid and reliable than classical cytogenetic method for the detection of cryptic Y-specific SRY sequence in patients with ambiguous genitalia.

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