University College London Hospitals NHS Foundation Trust

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Chromosomal variations in children and adolescents with gender dysphoria : is routine karyotyping indicated?

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Introduction

Gender dysphoria (GD) is a condition where an individual presents with extreme discomfort or distress due to an incongruity between their biological sexual characteristics assigned at birth and their gender identity.

The Manual of Mental Health Disorders (DSM V) (2) and its diagnostic criteria are used to form a formal diagnosis by the Tavistock and Portman in forming formal diagnosis of young adolescents presenting with significant dysphoria and enabling referral to endocrine liaison services for medical interventions.

Urgent karyotyping is always indicated in disorders of sex development (DSD), (1) However the need for chromosome analysis in gender dysphoria (GD) is less clear. We therefore reviewed its place in the management of this condition.

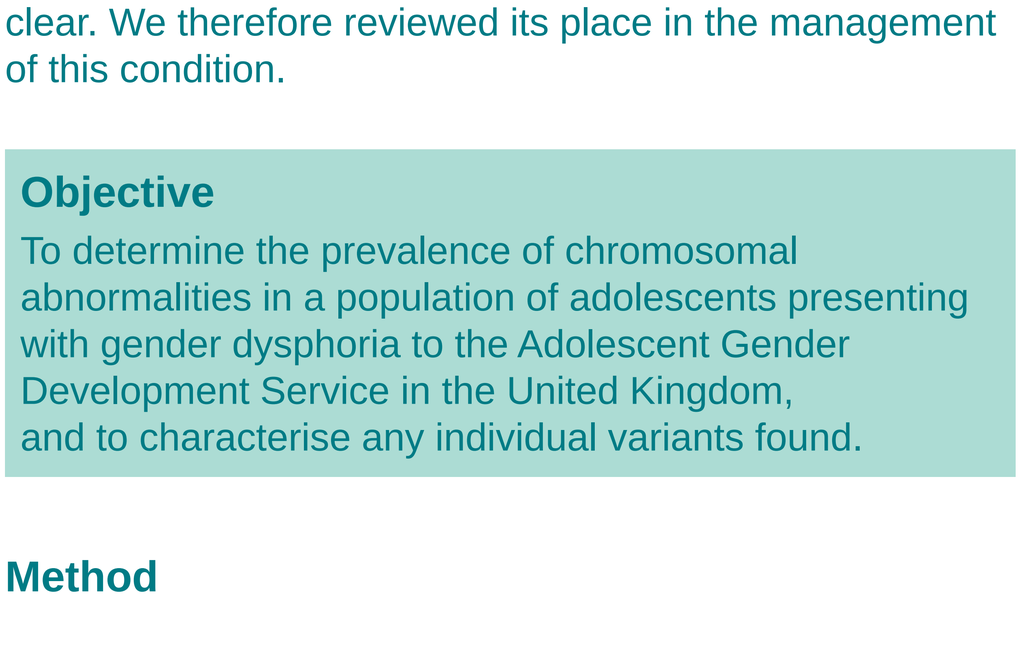
Objective

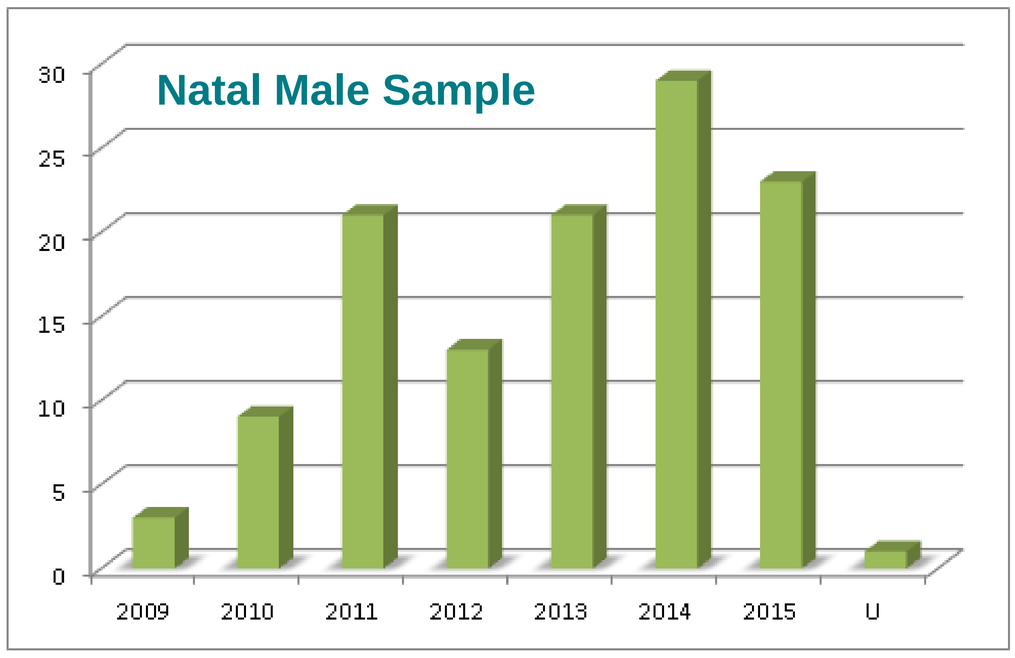
To determine the prevalence of chromosomal abnormalities in a population of adolescents presenting with gender dysphoria to the Adolescent Gender Development Service in the United Kingdom, and to characterise any individual variants found.

Method

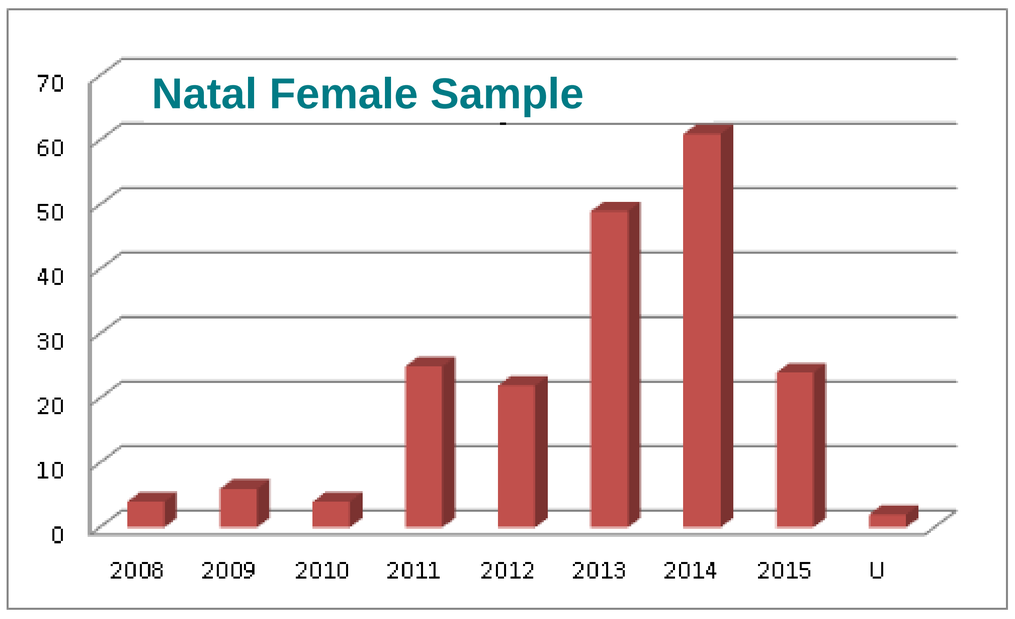
Chromosomes were collected from three hundred and seventeen patients; 197 female to males (FtM) and 120 male to female (MtF), with an average age of 16 years in both groups across a period of 8 years at the endocrine clinic at UCLH.

Results





Average age: 16	
2011	46,XYY



Average Age: 16	
	mos
2013	47,XX,+mar[10}/46,XX[20]
2008	46xx, t(7;13)(p21;q31)

One denovo sex chromosome variation (47,XYY) was identified. One prenatal diagnosis of 47,XXX was reconfirmed. Neither would have been suspected phenotypically nor did the finding have any bearing on management of the GD.

In addition, karyotyping revealed two individuals with balanced familial translocations and one person with a small marker chromosome, none of which had any clinical consequences. These karyotype variations are within known population aneuploidy prevalence rates (4)

Conclusion

No additional chromosome variations in children and adolescents with GD were identified over and above the frequency expected within the general population (3).This differs from the situation in a DSD. Chromosome analysis in children and adolescents with GD is therefore not routinely indicated.

References

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