Sex chromosome mosaicism (45,X/46,XY) is a rare subgroup of disorders of sex development. It has widespread clinical implications as it affects growth, hormonal balance, gonadal development, and histology. Clinical knowledge about the phenotype and outcome of this group of patients is limited.

The aim of this study is to evaluate phenotype, growth, reproductive hormones and gonadal histology in patients with 45,X/46,XY mosaicism.

This was a retrospective, longitudinal study conducted from January 2006 to January 2021 at a tertiary paediatric endocrine referral center. Patients’ clinical presentation, karyotypes, height, hormonal profiles, imaging and histologic findings of the gonads were evaluated. Phenotypes were scored using external masculinization scores (EMS) and external genitalia scores (EGS).

The 45,X/46,XY phenotype varies widely. This study demonstrates an association between the 45,X/46,XY karyotype and growth as has been described previously. Assessment of gonadal differentiation patterns in 45,X/46,XY is challenging because it represents a continuum between two extremes (normal testis and normal ovary). Management of each patient should be individualized according to the clinical and histological assessment.


2. Li Pan, Zhe Liu, Xiu Li, Longjiang Zhang, Shoul Li and on behalf of the Mosaicism Collaborative Group. The 45,X/46,XY phenotype varies widely. This study demonstrates an association between the 45,X/46,XY karyotype and growth as has been described previously. Assessment of gonadal differentiation patterns in 45,X/46,XY is challenging because it represents a continuum between two extremes (normal testis and normal ovary). Management of each patient should be individualized according to the clinical and histological assessment.

3. We thank all referring clinicians and patients who have contributed to this study.

4. The 45,X/46,XY phenotype varies widely. This study demonstrates an association between the 45,X/46,XY karyotype and growth as has been described previously. Assessment of gonadal differentiation patterns in 45,X/46,XY is challenging because it represents a continuum between two extremes (normal testis and normal ovary). Management of each patient should be individualized according to the clinical and histological assessment.

5. We thank all referring clinicians and patients who have contributed to this study.

6. The 45,X/46,XY phenotype varies widely. This study demonstrates an association between the 45,X/46,XY karyotype and growth as has been described previously. Assessment of gonadal differentiation patterns in 45,X/46,XY is challenging because it represents a continuum between two extremes (normal testis and normal ovary). Management of each patient should be individualized according to the clinical and histological assessment.

REFERENCES


2. Li Pan, Zhe Liu, Xiu Li, Longjiang Zhang, Shoul Li and on behalf of the Mosaicism Collaborative Group. The 45,X/46,XY phenotype varies widely. This study demonstrates an association between the 45,X/46,XY karyotype and growth as has been described previously. Assessment of gonadal differentiation patterns in 45,X/46,XY is challenging because it represents a continuum between two extremes (normal testis and normal ovary). Management of each patient should be individualized according to the clinical and histological assessment.

3. We thank all referring clinicians and patients who have contributed to this study.

4. The 45,X/46,XY phenotype varies widely. This study demonstrates an association between the 45,X/46,XY karyotype and growth as has been described previously. Assessment of gonadal differentiation patterns in 45,X/46,XY is challenging because it represents a continuum between two extremes (normal testis and normal ovary). Management of each patient should be individualized according to the clinical and histological assessment.

5. We thank all referring clinicians and patients who have contributed to this study.

6. The 45,X/46,XY phenotype varies widely. This study demonstrates an association between the 45,X/46,XY karyotype and growth as has been described previously. Assessment of gonadal differentiation patterns in 45,X/46,XY is challenging because it represents a continuum between two extremes (normal testis and normal ovary). Management of each patient should be individualized according to the clinical and histological assessment.