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Abstract Topic: - Clinical Genetics

Abstract Title: - Assessment of cytogenetic findings and risk factors in females with primary amenorrhea.

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Aims: - The present study aimed to explore the maximum factors responsible for primary amenorrhea including cytogenetics, radiology, and clinical profiles.

Methods: - A detailed proforma was filled with the informed consent of females with PA (cases) (n=45) and age-matched controls (n=45).

Results: - Results revealed body mass index (BMI) ($p<0.001$), stature ($p<0.001$), lifestyle factors ($p<0.05$), maternal factors ($p<0.001$), familial factors ($p<0.05$) and serum level of follicle-stimulating hormone (FSH) ($p<0.001$), luteinizing hormone (LH) ($p<0.001$), thyroid-stimulating hormone (TSH) ($p<0.05$), prolactin ($p<0.05$), estrogen ($p<0.001$), progesterone ($p<0.001$) and testosterone ($p<0.001$) were in significant association with the condition of PA in females. The cytogenetic analysis revealed 57.7% (n=26) PA females with normal karyotype (46,XX) suggesting Mullerian agenesis and 42.2% (n=19) females with abnormal karyotype including 45,X (n=3), 46,XY (n=4), mos 45,X[10]/46,XY[90] (n=1), mos 45,X[84]/47,XXX[16] (n=1), 46,X,del(X)(p22.2) (n=1), 46,X,i(X),(q10) (n=1), 46,XX,9qh+ (n=7) and 46,XX,9qh+,21ps+ (n=1). The development of secondary sexual characteristics (SSCs) and ovaries ($p<0.05$) in PA females was significantly associated with the cytogenetic findings.

Conclusions: - The present study provides insights regarding various indices responsible for the PA in females. Also, novel indices of maternal and family history provide the knowledge about the occurrence of condition and will help clinician to manage accordingly. BMI, height, maternal conception, number of pregnancies, birth order, birth weight, consanguinity, lifestyle habits, radiology profile, and hormone levels are all critical factors to be considered when providing counselling for managing the disease. Furthermore, conducting cytogenetic evaluation is essential for providing necessary psychological and management support to female patients and their families.

Keywords: - Results revealed body mass index (BMI) ($p<0.001$), stature ($p<0.001$), lifestyle factors ($p<0.05$), maternal factors ($p<0.001$), familial factors ($p<0.05$) and serum level of follicle-stimulating hormone (FSH) ($p<0.001$), luteinizing hormone (LH) ($p<0.001$), thyroid-stimulating hormone (TSH) ($p<0.05$), prolactin ($p<0.05$), estrogen ($p<0.001$), progesterone ($p<0.001$) and testosterone ($p<0.001$) were in significant association with the condition of PA in females. The cytogenetic analysis revealed 57.7% (n=26) PA females with normal karyotype (46,XX) suggesting Mullerian agenesis and 42.2% (n=19) females with abnormal karyotype including 45,X (n=3), 46,XY (n=4), mos 45,X[10]/46,XY[90] (n=1), mos 45,X[84]/47,XXX[16] (n=1), 46,X,del(X)(p22.2) (n=1), 46,X,i(X),(q10) (n=1), 46,XX,9qh+ (n=7) and 46,XX,9qh+,21ps+ (n=1). The development of secondary sexual characteristics (SSCs) and ovaries ($p<0.05$) in PA females was significantly associated with the cytogenetic findings.