# PHENOTYPE/ GENOTYPE CORRELATION IN FEMALES WITH GONDAL DYSGENESIS

## مقارنة الشكل الظاهرى بالتركيب الوراثى فى الإناث المصابة بالخلل التكوينى فى الغدد الجنسية

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الدكتوراه
فى الوراثة


**By**
**Mona Kamal Mekkawy**
**M.B., B.Ch.**
**(Ain Shams University)**
**M.Sc. Pediatrics ( Ain Shams University)**
**Assistant Researcher**
**Human Genetics Dept.**
**National Research Center**
**Cairo**

من
منى محمد كمال مكاوى
بكالوريوس الطب والجراحة
(جامعة عين شمس)
ماجستير طب الأطفال
(جامعة عين شمس)
باحث مساعد
المركز القومى للبحوث القاهرة
قسم الوراثة البشرية
القاهرة


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## Supervisors

**Professor Doctor**

**Suzan Roshdy Ismail**

**Professor of Human Genetics**

**Medical Research Institute**

**Alexandria University**

**Professor Doctor**

**Samia Ali Temtamy**

**Professor of Human Genetics**

**National Research Center**

## Co-Supervisors

**Professor Doctor**

**Ibtessam Ramsy Hussein**

**Professor of Human Genetics**

**National Research Center**

**Doctor**

**Nargues Mahmoud Hassanein**

**Assistant Professor of Human Genetics**

**Medical Research Institute**

**Alexandria University**

## المشرفون

الأستاذ الدكتور

**سوزان رشدى إسماعيل**

أستاذ الوراثة البشرية

معهد البحوث الطبيية

جامعة الإسكندرية

الأستاذ الدكتور

**سامية على التمتامى**

أستاذ الوراثة البشرية

المركز القومى للبحوث القاهرة

المشرفون المشاركون

الأستاذ الدكتـــور

**ابتسام رمزى حسين**

أستاذ الوراثة البشرية

المركز القومى للبحوث القاهرة

دكتـــورة

**نرجس محمود حسنين**

أستاذ مساعد الوراثة البشرية

معهد البحوث الطبية

جامعة الإسكندرية

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# List of Abbreviations

| | |
|---|---|
| **AHC (DAX 1)** | Adrenal Hypoplasia Congenital. |
| **AMH** | Anti-Mullerian Hormone. |
| **AZF** | Azoospermia Factor(s). |
| **BMP15** | Bone Morphogenetic Protein 15 gene. |
| **CD** | Campomelic Dysplasia. |
| **CMPD** | Campomelic Dysplasia locus. |
| **DAZ** | Deleted in Azoospermia. |
| **DDS (DAX 1)** | Dose – Dependent Sex reversal. |
| **DFFRX** | Drosophila Fat Facets Related X. |
| **DHT** | Dihydrotestosterone. |
| **DIAPH2** | A human homologue of Drosophila Diaphanous (dia) gene. |
| **DMRT** | Double sex and Mabs Related Transcription factor. |
| **DNA** | Deoxy – Ribonucleic Acid. |
| **DSD** | Disorders of Sex Development. |
| **FISH** | Fluorescent In Situ Hybridization. |
| **FMR** | Fragile X Mental Retardation. |
| **FSH** | Follicle Stimulating Hormone. |
| **GBY** | Gonadoblastoma Y locus. |
| **GD** | Gonadal Dysgenesis. |
| **HCG** | Human Chorionic Gonadotropin. |
| **HMG** | The main functional DNA- binding domain of the SRY protein. |
| **IQ** | Intelligence Quotient |
| **LH** | Lutenizing Hormone. |
| **MIS** | Mullerian Inhibitory Substance. |

# List of Abbreviations (cont.)

| | |
|---|---|
| **MPH** | Male Pseudo-Hermaphroditism. |
| **MR** | Mental Retardation. |
| **PAR 1** | Pseudo-Autosomal Region 1 |
| **PAR 2** | Pseudo-Autosomal Region 2 |
| **SRA** | Autosomal Sex Reversal locus. |
| **SRY** | Sex-determining Region of the Y chromosome. |
| **SF-1** | Steroidogenic Factor-1 |
| **SOX 9** | A member of SOX ( SRY- related HMG box gene family). |
| **TDF** | Testis Determining Factor. |
| **TSPY** | A multicopy gene located in interval 3 on Y long arm (normally expressed in spermatogonial cells of the testes). |
| **UBE 1** | Ubiquitation Pathway Enzyme. |
| **USP9X** | Ubiquitin – Specific Protease 9 |
| **WT-1** | Wilms' tumor suppressor gene. |
| **ZFX** | Zinc Finger – X. |
| **ZFY** | Zinc Finger – Y. |

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# Introduction

# Review of Literature

# Patients and Methods

# Results

# Discussion

# Summary