

**Evaluation of Genetic Counseling for
Cases referred to Genetic Unit,
Pediatric Hospital, Ain Shams
University**

Thesis

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List of abbreviation

<i>Abbr.</i>	<i>Full term</i>
AAD	Amino acid disorder
ALPS	Auto immune lymphoproliferative disorder
CAPMAS	Central Agency for Public Mobilization Statistics
CHD	Congenital heart disease
CP	Cerebral palsy
CT	Computed topography
CVS	Chorionic villous sampling
DM	Diabetes mellitus
EDD	Estimated date of delivery
FU	Follow up
GC	Genetic counseling
GDD	Global developmental delay
HTN	Hypertension
I.E.M	Inborn errors of metabolism
LSD	Lysosomal storage disorder
MEN	Multiple endocrine neoplasia
MRI	Magnetic resonance imaging
MS	Multiple Sclerosis
NICU	Neonatal intensive care unit
NMD	Neuromuscular disorder

OA	Organic acidemia
PICU	Pediatric intensive care unit
PKU	Phenyl ketonuria
SD	Standard of deviation
SES	Socioeconomic status
Sib	Sibling
TORSCH	Toxoplasma, Rubella, Syphilis, CMV and HSV infection
UCD	Urea cycle disorder
US	Ultrasound sonography
WHO	World Health Organization

ABSTRACT

To direct effective preventive, diagnostic and counseling services, collecting data on the incidence and prevalence of various congenital and genetic disorders and their risk factors is a pre-requisite for establishing genetic services at the community level and mainly at the primary health care setting. The current work is a cross sectional study on patients attending the genetic clinic, Ain shams university. It is a trial to identify the characteristics of couples attending the clinic for genetic counseling and to assess the success of the available investigations in reaching the definitive diagnosis for proper counseling is important targets