

# INSTITUTE OF HUMAN GENETICS

ISO 9000:2000

GENETICS CENTRE

Reg. No. : 648

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**Report On :**

**Delivery : By Mail :**

**Personal :**

## PROFORMA

Ref. No.

Date :

Name :

Age : Sex : M / F

D.O.B. :

Father's / Husband's Name :

Age : Occupation :

Mother's Name :

Age :

Address for Correspondence :

Phone No. :

Origin From :

Caste :

Referring Hospital and/or Name of the Doctor :

Investigation From : Blood / Abortus / Amniotic / CVS / Bone Marrow / DNA / FISH

*Indication For Study :*

- a) Bad Obstetric History
- b) Multiple Congenital Anomalies
- c) Abnormal Sexual Development
- d) Psychomotor Retardation
- e) Down Syndrome / Triple Marker Positive
- f) H/o. Genetic Defects in the family
- g) Skeletal Deformities
- h) Leukemia AML, CML, MDS, MPDS

(2)

Obstetric History : Gr \_\_\_\_\_ Para \_\_\_\_\_ Ab \_\_\_\_\_

Still Birth \_\_\_\_\_ Twins \_\_\_\_\_

Pregnancy : Uncomplicated / / Complicated by Chronic Illness / /

Toxemia / / Trauma / / Drugs / / O.C. / Hormones /

Radiation / / Viral Infections / / Bleeding / /

Labour : Fetal Distress / / Diminished Fetal Movements / / Normal /

Prolonged / / Other Complications : \_\_\_\_\_

Delivery : Hospital / Home : Full Term / / Premature / / Post Mature

/ / Normal / / Instrumentation / / Asphyxia /

Cyanosis / / Cried soon after birth / /

Neonatal : At birth : Weight \_\_\_\_\_ Length \_\_\_\_\_ H.C. \_\_\_\_\_

Feeding Breast top wt-gain : Good / Fair / Poor

Convulsions :

Jaundice :

Milestones : Smiling- Head Holding- Recognizing- Turning Over-

Sitting- Standing- Walking- Talking-

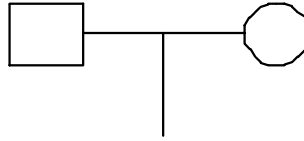
Toilet Training-

Family History : Mental Subnormality  Diabetes, Hypertension

Congenital Malformation  Consanguinity

(3)

**PEDIGREE**



Reports

Blood Counts :

Bone Marrow Counts :

Folate : B12 :

HCY :

Torch : aPL :

VDRL :

ANA :

USG Findings : Nuchal Fold

Triple Marker Results: AFP :

HCG :

UE3 :

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**CONSENT FORM**

**CONSENT TO CARRY OUT CHROMOSOME STUDY FROM :**

**CHORIONIC VILLI / AMNIOTIC FLUID / ABORTUS MATERIAL / BONE MARROW / BLOOD :**

I/we \_\_\_\_\_ give my/our consent to carry out Genetic Study at Genetics Centre, FRIGE House, Satellite, as a diagnostic test. I/We fully agree and understand that under any circumstances our culture may fail necessitating recollection of the sample, if possible. I/we understand that even under normal circumstances our cells may not be able to grow making our genetics study difficult.

I/We therefore agree to give another sample if needed.

It is further agreed that the nature of this agreement is such that it must remain confidential and we agree that the sole copy of the agreement may be retained in the above doctors file and shall not be disclosed except under unavoidable circumstances.

In case of paternity test we agree that any legal matter arising out of this, the Genetics Centre shall not be liable for this.

For any court appearance if needed, it will be at the cost of the patient.

Our centre will not be responsible for any dispute arising out of the DNA result.

Ref. No. :

Date :

Signatures :

Relation :

Time :

Name :

Place :

Witness : 1

(4)

*Physical Examination :*

*Height : Weight : Head Circumference : Span :*

*Skull : Shape : Flattening : Prominence :*

Fontanelles: Anterior : Posterior :

Statures : Prominent : Separated / Normal :

Hair : Normal /        / Thin /        / Sparse /        / Brittle /        /

*Facies : Flat / Mongoloid Cretenoid Asymmetry*

*Eyes : N / Small / Large                      Strabismus                      Nystagmus*

Slant N / Up / Down Pupils Cornea

Epicanthic folds

*Ears : N / Small / Large Shape : Rotation : N / Ant Post*

Position N / Lowest Malformation

*Nose : N / Large*

Bridge : Depressed / Hypertelorism

*Mouth : Cleft lip / Cleft Palate Tongue : Protruding / Thick*

Palate : High Arched / Narrow Micrognathia / Retrognathia

*Neck : Short / Low hair line / Webbing / Excess Fat*

*Cardiac Problems :*

*Limbs : Hands Short / Long / Normal Figures : Short. Stubby / Long thin / Non*

Clinodactyly /        / Polydactyly /        / Loops /        / Whirls /

Arches /        / Dermatoglyphics : Simian crease : Unilateral / Bilateral /

Bridge / Absent Proximal 'T' (ATD angle) : N / Distally placed

Foot : Planter Furrow /        / Increased space between 1st & 2nd toe /

Syndactyly /        /

*Joints : Normal /        / Stiff /        / Contractures /        / Dislocation /*

*Appendages : Nails-N / Hypoplastic / Thick*

Hair-N / Hirsutism / Kinky / Brittle

Skin-N / Rough / Infection

*Genitals : Hypospadias Cryptorchidism*

*Normal Male / Female*

*Others*

(5)

*Investigation and Follow-up :*

- A. X or Y Chromation
- B. Karyotype
- C. Fibroblast Culture
- D. Gonadal biopsy-Meiotic studies
- E. Biochemical screening
- F. Amniocentesis
- G. Family Studies
- H. Hormones

I. Triple Marker Screening : AFP :

BHCG :

UE3 :

J. DNA Report : Father :

Mother :

Proband :

Child :

CVS :

AF :