



Tehran University of Medical Sciences

# Medical Genetic Services as a Health Tourism Attraction

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ربنا هب لنا من ازواجنا وذريتنا قره اعين و جعلنا للمتقين اماما



## ➤ Iran Health Tourism Attractions

**different fields;**

**Cardiac surgery**

**Plastic surgery**

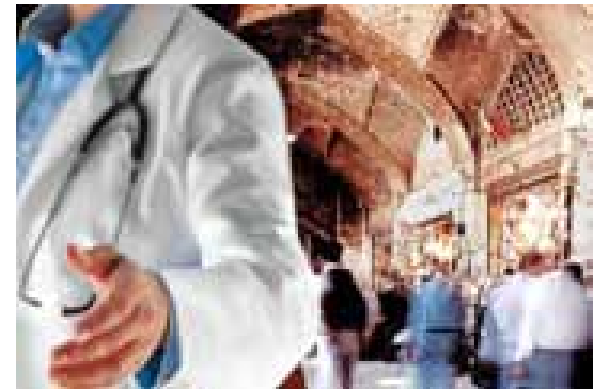
**Ophthalmology**

**Infertility**

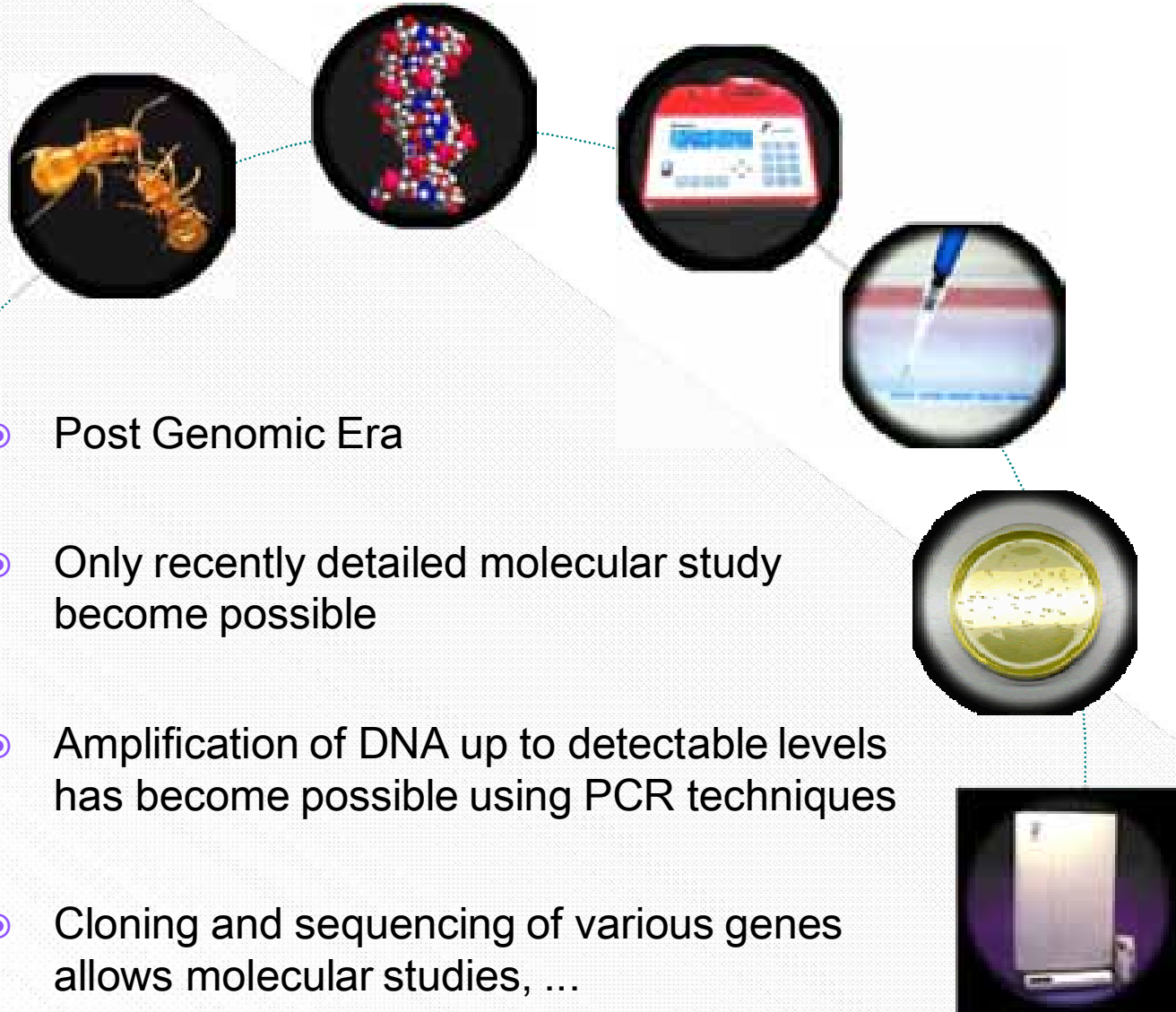
**Organ transplantation**

**Medical Genetics**

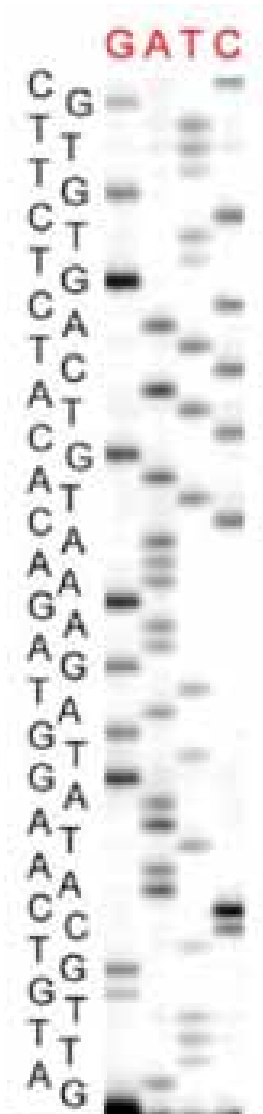
...



# The DNA revolution

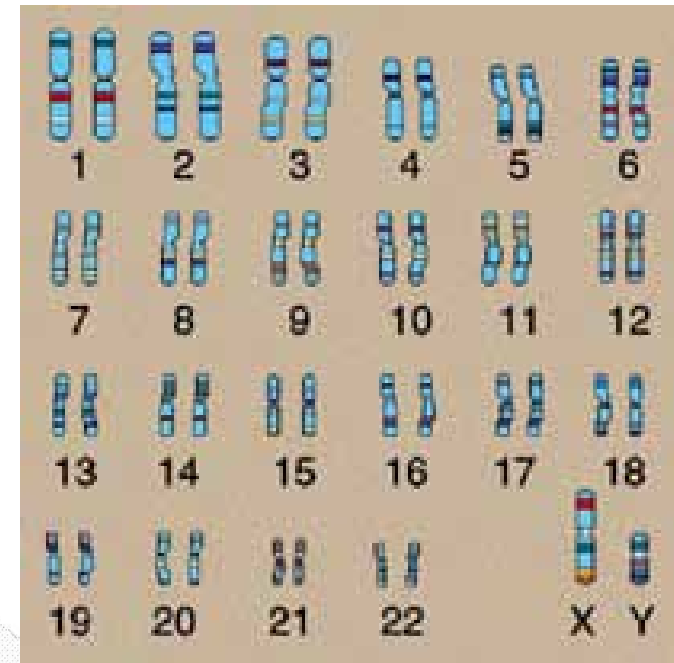


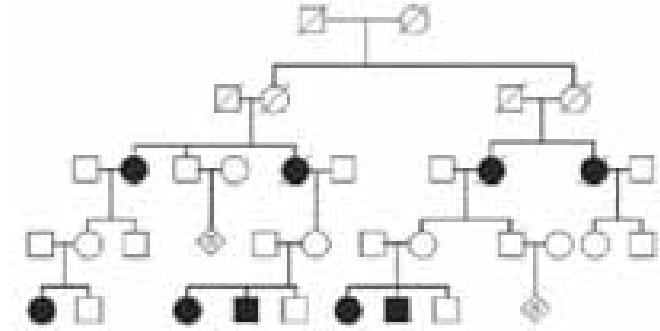
- Post Genomic Era
- Only recently detailed molecular study become possible
- Amplification of DNA up to detectable levels has become possible using PCR techniques
- Cloning and sequencing of various genes allows molecular studies, ...



# Genetic Determinants of Disease

- **Chromosomal abnormalities**
  - > Loss or gain of chromosomes
  - > Loss or gain of chromosomal regions
  - > Chromosomal translocation
- **Single gene disorders**
  - > Coding regions – loss, gain, or alteration of protein function
  - > Regulatory regions – increased, decreased or inappropriate expression
- **Multifactorial disorders**
  - > Gene-Environment interactions



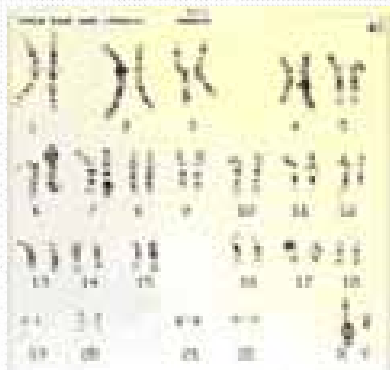


مشاوره ژنتیک دقیق و مشروح، آزمایشات کروموزومی و مولکولی

ابزارهای رویکرد ژنتیک در بیماری ها

Detailed Genetic Counseling, Molecular and cytogenetic studies

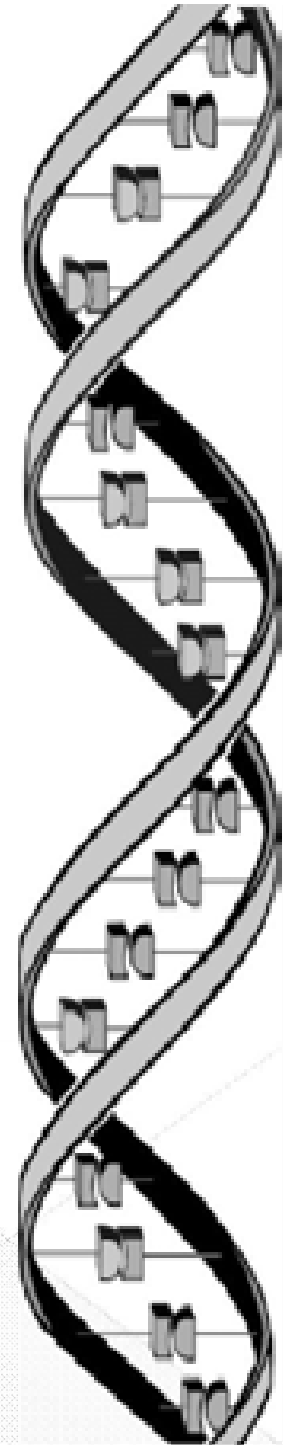
Tools for medical genetic approach



Case detail;  
Genetic Counseling

Cytogenetic study; Karyotyping  
Molecular study;  
SSCP, PCR/ RFLP, paternity test

PND, PGD





سید  
مفتی سید محمد تقی حسینی  
العلیہ السلام

۵۹۰۲۴

نام معلم و مدرس  
مدرسہ  
نام مدرسہ

اشتراک اینک با مکتبہ شریعتیه علامہ علی قاری در نشر کتاب و کتب  
مکتبہ جامعہ و کتب مکتبہ جامعہ

شماره ۱۰۰ یا ۱۰۱ با کتب مکتبہ جامعہ در نشر کتاب و کتب مکتبہ جامعہ

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۸۱، ۵، ۲۷

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دفتر اعلیٰ

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۷۵، ۳، ۲۴

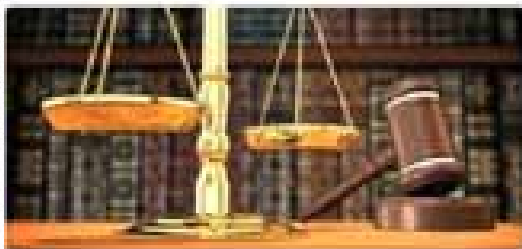


## act of “Therapeutic Abortion”

Approved in 2005  
by Parliament and Guardian Council;

“TA is permissible with definite diagnosis of 3 specialist clinicians and approval of legal medicine due to fetus disorders which is a big problem for mother based on retardation or anomaly and or mother disorders threatening her life before ensoulment (4 months of gestation) with consent of mother and there is no penalty for physician performing such service.

Violators of this act would be punished based on Islamic punishment law”.



## قانون سقط درمانی

ماده واحده – سقط درمانی با تشخیص قطعی سه پزشک متخصص و تایید

پزشکی قانونی مبنی بر بیماری جنین که به علت عقب افتادگی یا ناقص

الخلقه بودن موجب حرج مادر است و یا بیماری مادر که با تهدید جانی

مادر توأم باشد قبل از ولوج روح (چهار ماه) با رضایت زن مجاز می باشد

و مجازات و مسئولیتی متوجه پزشک مباشر نخواهد بود.

متخلفین از اجرای مفاد این قانون به مجازات های مقرر در قانون مجازات

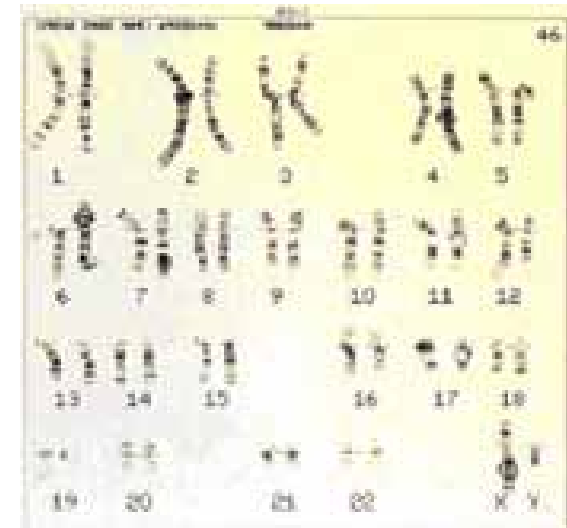
اسلامی محکوم خواهند شد.

قانون مصوب مجلس شورای اسلامی ۱۳۸۴/۳/۱۰

تایید شورای نگهبان ۱۳۸۴/۳/۲۵

# *Populations at Risk for Chromosome Errors*

- spontaneous recurrent abortion
- sexually ambiguous
- infertile males or females
- newborns with multiple congenital anomalies
- Growth, development or mentally retarded



## *Indications for Prenatal diagnosis (PND)*

- Advanced maternal age
- Child with chromosomal abnormality
- Either partner is carrier of chromosomal aberration
- History of known hereditary disorder for which detection is possible and has been established in family
- Unusual maternal screening markers
- Exposure to teratogen agent

# *Prenatal Diagnosis Techniques*

- **Visualization:** Sonography  
Radiology
- **Fetal Samples:** Amniocentesis (13-20 wk.)  
Chorionic villi sampling (10-12 wk.)
- **Fetal Screening:**

|  |           |
|--|-----------|
| 3M: $\alpha$ FP, $\beta$ hcG, estriol            | 15-20 Wks |
| 1 <sup>st</sup> trimester: NT, PAPP, $\beta$ hcG |           |



continent



country



city



building



cell nucleus

chromosome

chromosome band

gene

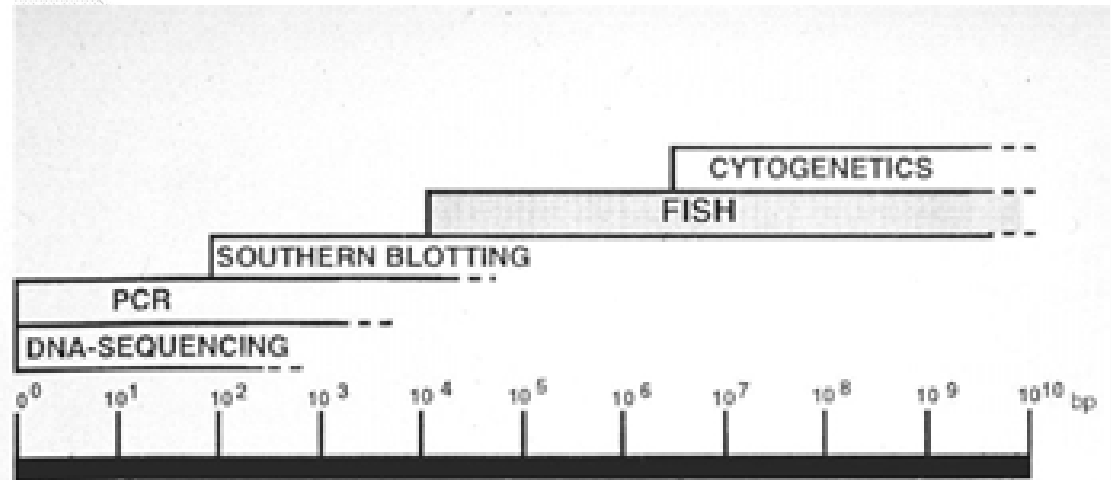
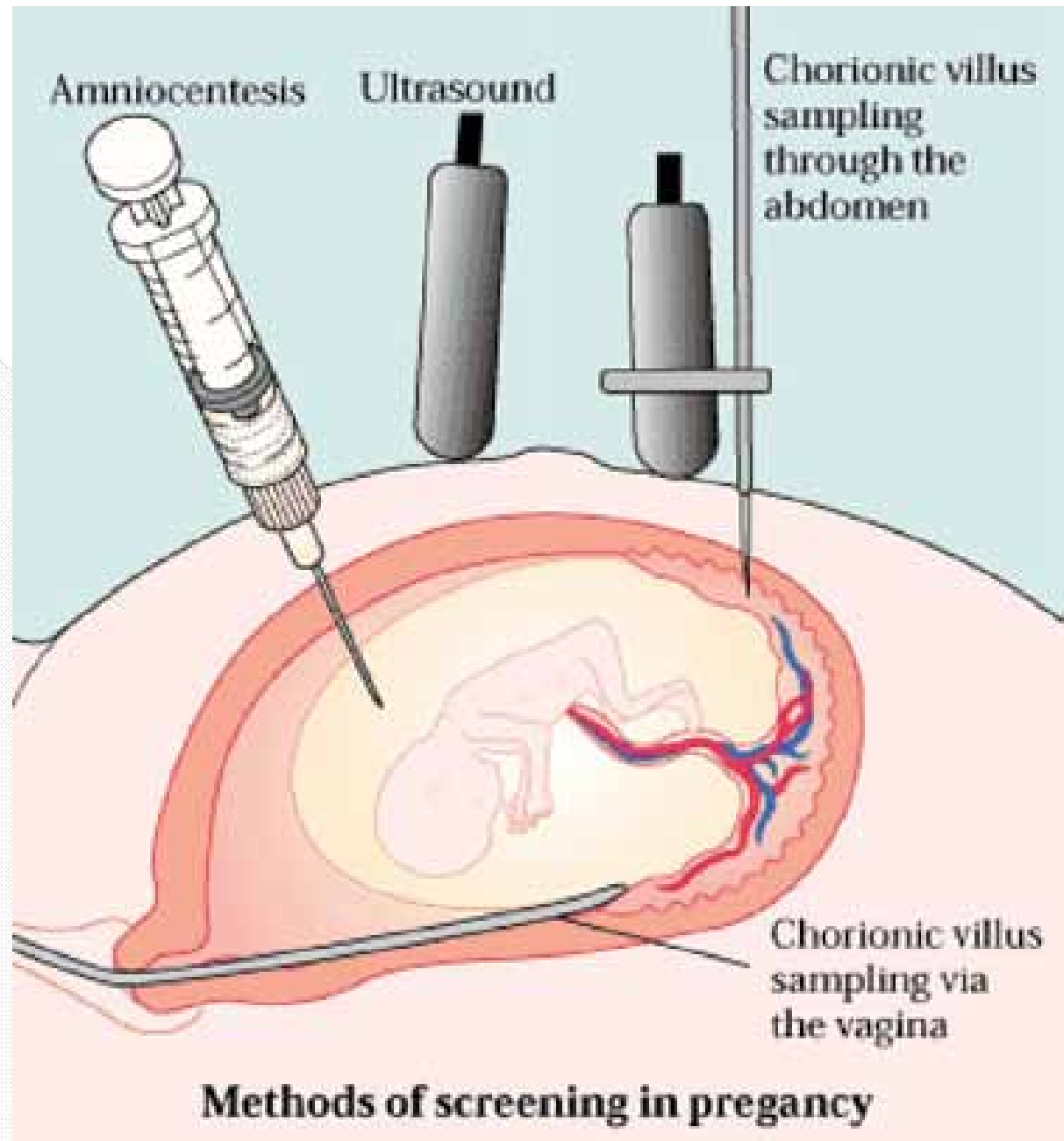


Fig. 6. The power of resolution using standard genetic techniques for detection of mutations.

# Prenatal diagnosis (PND)

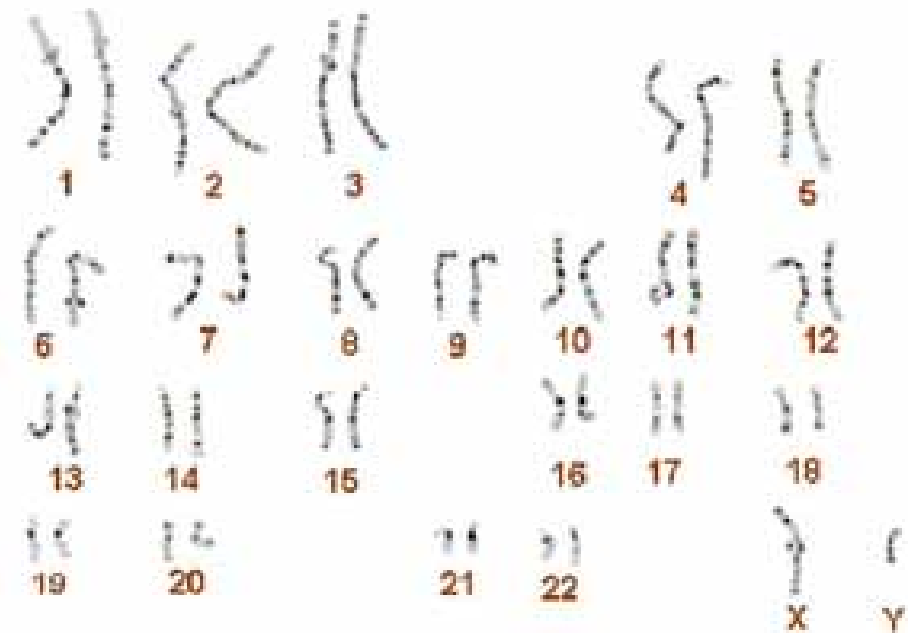


# Examining chromosomes



Metaphase spread

karyotype



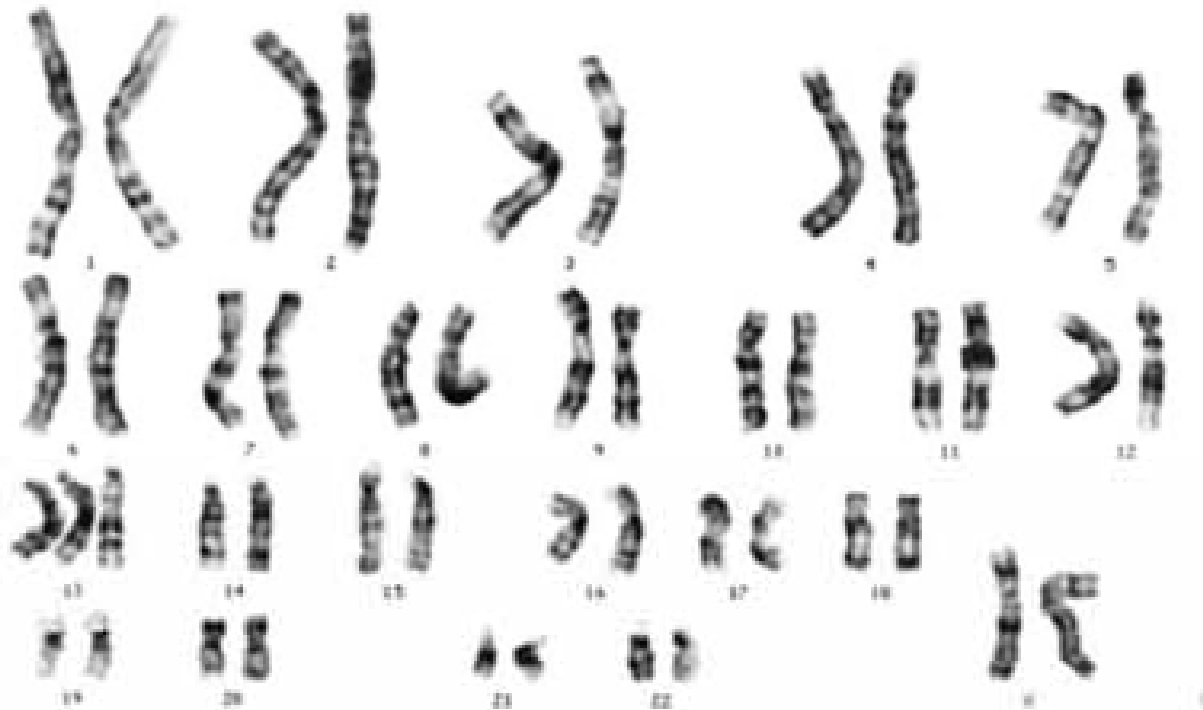


Trisomy 21 (Down syndrom)





# Trisomy 13

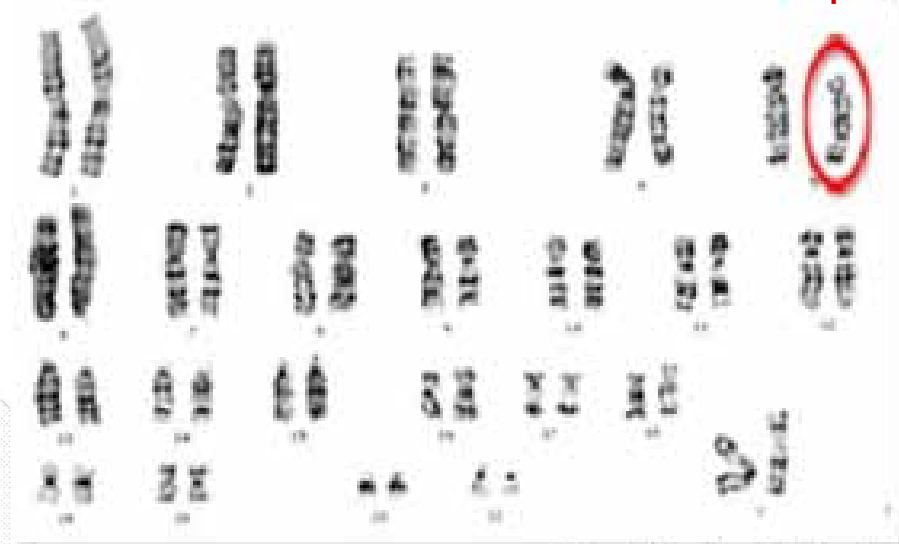


- Cleft lip/palate
- Polydactyly (postaxial)
- Microcephaly
- Omphalocele
- Scalp defect

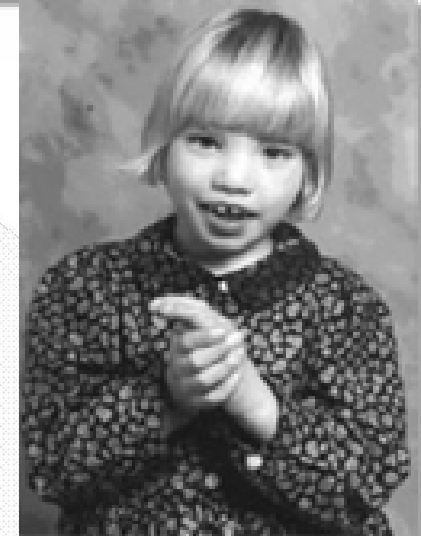


# Cri du chat syndrome

5p-



Cat cry, hypertelorism, epicanthus, mental retardation



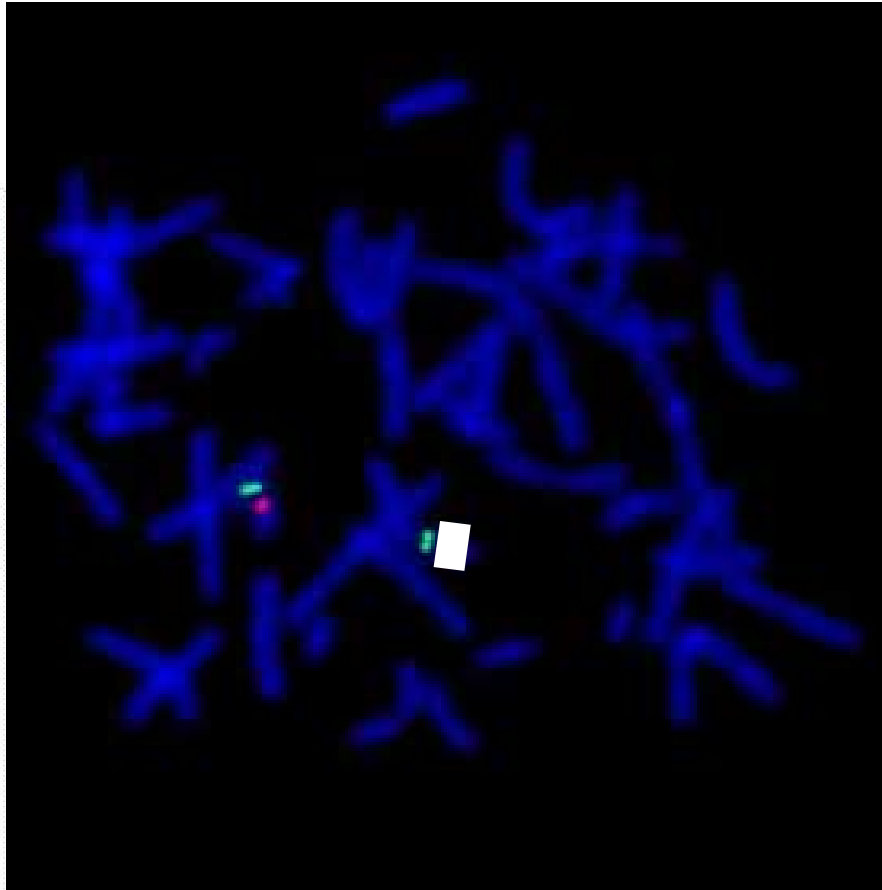
CdC Support Group  
[www.cricchat.u-net.com](http://www.cricchat.u-net.com)

# DiGeorge syndrome



C: cardiac  
A: anomaly  
T: thymus  
C: cleft lip  
H: hypocalcemi

CATCH 22



# Emerging Technologies

## Preimplantation Genetic Diagnosis (PGD)

- Use of molecular or cytogenetic techniques during in vitro fertilization (IVF) to select embryos free of a specific genetic condition for transfer to the uterus.
- This technology was developed to offer an option to couples at significant risk for a specific genetic disorder but who are opposed to abortion
- is performed on a single cell from a 6-8 cell blastomere or on a polar body after IVF .

# PGD

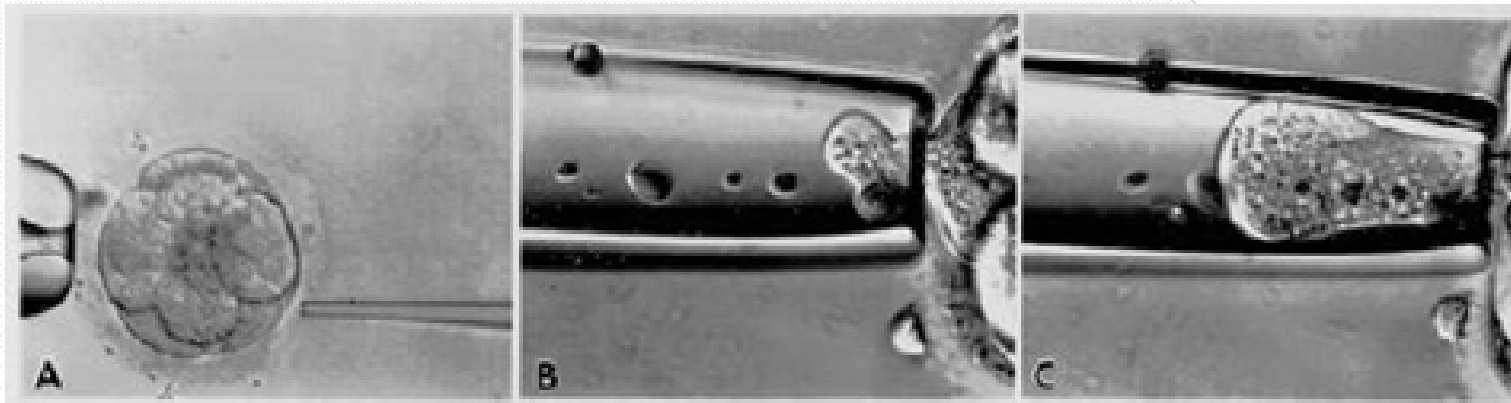
## Timeframe: 3 days post-insemination

embryos obtained through in vitro fertilization

one or two cells are removed from the 6-10 cell blastomere

cells are analysed by FISH (for chromosomal abnormalities) or PCR  
(for mutation analysis)

selected embryos are transferred at the end of day 3



# different aspects of consanguinity

## 1) Problem?

AR disorders  
Congenital anomaly

## 2) Preferred?

- a) Strengths
- b) Keep the wealth within family
- c) Support women from men family side & vice versa

*J. Inher. Metab. Dis.* page 1 of 4, © 2006 Cambridge University Press  
doi:10.1017/S0021962306001444

### Short Report

## IS CONSANGUINEOUS MARRIAGE RELIGIOUSLY ENCOURAGED? ISLAMIC AND IRANIAN CONSIDERATIONS

SEYED MOHAMMAD AKRAMI\*† and ZAHRA OSATI\*

\*Medical Genetics Department and †Endocrinology and Metabolism Research Center,  
Tehran University of Medical Sciences (TUMS), Tehran, Iran

# Future of genetic disorders in our region?

J Genet Counsell (2015) 15:42–50  
DOI 10.1007/s10897-014-9781-z

ORIGINAL RESEARCH

## Is There a Significant Trend in Prevalence of Consanguineous Marriage in Tehran? A Review of Three Generations

Seyed Mohammad Akrami · Vahideh Mostafaei ·  
Samaneh Bahadi Shamsi · Bahareh Dehghan ·  
Bahar Lotfzad

Journal of Pediatric Genetics 1 (2012) 207–220  
DOI: 10.12139920.12014  
KJVS Press

## Review Article

# Genetics of consanguineous marriage: Impact and importance of counseling

Seyed Mohammad Akrami\*

Department of Medical Genetics, Tehran University of Medical Sciences, Tehran, Iran

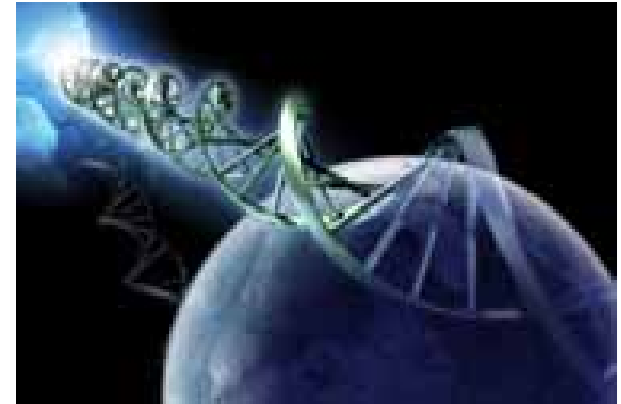
Table 1  
consin marriage in some of Middle East countries

| Country      | Percentage | Reference no |
|--------------|------------|--------------|
| Iraq         | 29         | 15           |
| Kuwait       | 30         | 16           |
| Saudi Arabia | 26         | 17           |
| Oman         | 24         | 18           |
| Jordan       | 32         | 19           |
| Turkey       | 22         | 20           |
| Afghanistan  | 46         | 27           |
| Syria        | 35         | 28           |

## PhD in Medical Genetics in 3 universities



Tehran University of Medical Sciences



Shahid Beheshti University of Medical Sciences



دانشگاه علوم بهزیاری و توانبخشی

University of Social Welfare And Rehabilitation  
Sciences

## MSc in Human Genetics in 13 universities





## Further reading

N Engl J Med. 347(23)  
Dec 2002

GENOMIC MEDICINE

Review Article

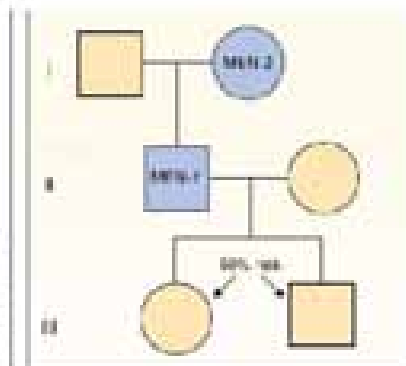
### Genomic Medicine

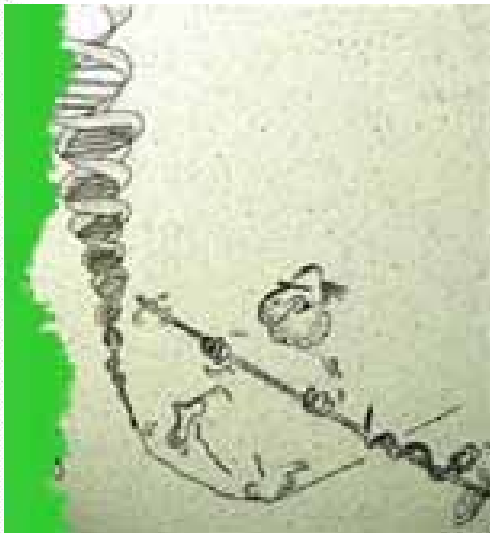
Alan E. Guttmacher, M.D.,  
and Francis S. Collins, M.D., Ph.D. (Pages 27-36)

#### GENETIC TESTING

Wen Han, M.D., A.S.

**G**ENETIC testing can provide concrete clinical benefits. A child known to have multiple sclerosis (MS) can be spared medication side effects by undergoing prophylactic thymectomy (Fig. 1) and an adult with hereditary hemochromatosis can be spared cirrhosis by the early initiation of phlebotomy therapy. Genetic testing can also reveal disease risk and prognosis, alter





Thank you for your  
kind attention

با تشکر از بذل توجه شما

