



PRECONCEPTION DIAGNOSIS

A challenge to prevent genetic diseases and handicap

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This publication, conceived to widen the knowledge of pathologies which can be related to reproduction, is addressed to couples who wish to engage in pregnancy, and it wishes to be a safe guide in this adventurous but exciting journey.

The work summarises various risk situations during pregnancy and suggests the necessary exams for an early determination of all those couple pathologies, hereditary or acquired, which can have negative consequences on the baby wished to be conceived.

Dedicated...

To all parents who did not have the chance to "prevent" and to their babies with "broken wings", so that their suffering will become the infallible motive for us researchers, clinicians and good-willing people, to win the challenge against time for the health and the happiness of tomorrow's children and families.

Exams and diagnostics: few Italian legislative aspects

by Carlo Giacobini

The diagnostic protocol for the protection of the responsible maternity (attachment A of the decree) is applied in the preconception phase and it foresees the exemption of the participation to the expenses for certain services, such as the verification of the blood group and RH factor; specific tests for the congenital blood diseases; HIV antibodies; colposcopy; smear-test. Furthermore, if some risk situations caused by hereditary pathologies arise from the anamnestic investigation or from the clinical conditions of the couple, upon request of the specialist or the geneticist, the same exemption can be applied to some diagnostic services such as karyotype, DNA analysis and test for congenital enzyme diseases. From a practical point of view, major difficulties are indicated, in these cases, to obtain the authorization to mate such exams in centres outside the local sanitary unit. The second diagnostic protocol (attachment B) lists the exams which do not forecast a participation to the expenses during pregnancy; the attachment foresees for each month of gestation, specific clinical analyses charged to the National Health Service. Again it is established that if "from the anamnestic investigation or from the clinical conditions of the pregnant woman arise risk situations caused by hereditary pathologies or related to the advanced age of the mother, certified by the specialist and/or the geneticist, the exemption of the participation to the expenses is extended to the exams finalized to the early prenatal diagnosis, to be done within the first three months or afterwards (amniocentesis). In theory you can note that some legislative assumption can contribute to the achievement of the objectives of prevention. But we must say that this achievement depends more from the service organization than from the procedures of the expenses exemption.

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To all parents who did not have the chance to "prevent" and to their babies with "broken wings", so fiat their suffering will become fie infallible motive for us researchers, clinicians and good willing people, to win the challenge aganst time for the health and fie happiness of tomorrow's children and families.

Some of the Italian centres for the genetic consultancy and fetal diagnosis

MILAN

Istituto "L. Mangiagalli", Prima Clinica Ostetrica e Ginecologica, Universita di Milano, Via Commenda 12 - PH.+39 0257992321

MILAN

Ospedale "San Raf&de", IRCCS, Laborato rio di Biologia Molecolare, Via Olgettina - Ph.+39 0226431

GENOA

Divisione Ostetrica, Istituto "G. Gaslini", Via 5 Maggio 39, Ph.+39010 56361

TURIN

Istituto di Gaietica Medica, Universita di Torino, Via Santena 19 - Ph. +39011 6336771

BOLOGNA

Policlinico S. Orsola, Cattedra di Fisiopatologia Prenatale, Via Massarenti 13 - Ph. +39 051 6363610

TRIESTE

Ospedale Burlo Garoflto, Istituto di Genetica Medica, Universita di Trieste, Via dell 'Istria 65 - Ph. +39040 3785424

FLORENCE

Policlinico di Careggi, Istituto di Clinica Ostetrica e Ginecologica, Univereita di Firenze, Viale Morgagni - Ph. +39 0554271421

PISA

Clinica Ostetrica e Ginecologica, Universita di Pisa, Via Roma 35, Centra di Medicina Preventiva per la Riproduzione (Dr. Strigini - Ph.+39050992530).

Clinica Pediatrica, Consulenza Genetica Prenatale fDr. Tarantino - Ph. +39 050 992139), Laboratorio di genetica (Dr. Sirai - Ph. +39050992644)

PISA

Istituto Scientifico "Stella Mans", Laboratorio di Neurogenetica e Dismorfblogia, Viale del Tineno 331 (Calambrone-Pisa)- Ph. +39 050 886111

ROME

Policlinico Gemelli, Istituto di Genetica Umana, Univeisita Cattolica del Sacra Cuore, Largo F. "Vote 1 -Hi.+390630151

CAGUARI

Ospedale Regionale per le MScrocitemie, Diagnosi Prenatale, Via Jenner - Ph. +39 070 6095542

PALERMO

Ospedale "V. Cervello", Centra di Diagnosi Prenatale, Via Trabucco 180 -Ph. +39 091 6802684

ENNA

Laboratorio di Diagnosi Genetica, IRCCS, Oasi di Troina - Ph.: +39 0935 936111

PA THOGENIC NOXAE

responsible for miscarriage, embryo and fetus pathologies

INFECTIOUS DISEASES

(rubeola, viral hepatitis, HIV, toxoplasma, citomegalovirus, herpes virus, Lue)

ENDOCRINE-METABOLIC DISEASES

(diabetes, thyroid disorders)

DRUGS

(thalidomide, anticoagulants, oral antidiabetics, sulpha drugs, streptomycin, etc.)

DEFICIENCIES OR MATERNAL NUTRITIONAL EXCESSES

ALCOHOL

CHEMICAL COMPOUNDS

(Lead, Mercury, Adhesives)

STEROID HORMONES

(diethylsilbestrol and anabolic steroids)

DRUGS

(heroin, methadone, cocaine, LSD, and as consequence abstinence crisis, vasoconstriction)

ACTIVE AND PASSIVE SMOKE

ENVIRONMENTAL FACTORS

(combustion products' benzopyrene, aromatic hydrocarbons)

RADIATION

(ionogenic radiations, electro-magnetic pollution)

PRECONCEPTION SCREENING
FOR HEALTHY COUPLES

Woman

- BLOOD CHEMISTRY + GROUP AND RH
FACTOR

- COOMBS INDIRECT TEST (*irregular antibodies identification*)
- ELECTROPHORESIS Hb (*pathologic Hb research*)
- POSTPRANDIAL GLYCEMIA (*to reveal hidden or potential diabetes*)
- RUBEO TEST
- TOXO TEST
- CITOMEGALOVIRUS + HERPES VIRUS
- VDRL e TPHA (*syphilis*)
- HIV (*immunodeficiency*), HCV, HBSAg (*hepatitis C and B*)
- VAGINAL TAMPON (HPV)
- SMEAR-TEST and COLPOSCOPY

Man

- BLOOD CHEMISTRY + GROUP AND RH FACTOR

- ELECTROPHORESIS Hb (*pathologic Hb research*)
- CITOMEGALOVIRUS + HERPES VIRUS
- HIV (*immunodeficiency*), HCV, HBSAg (*hepatitis C and B*)

PRECONCEPTION

FOR PREGNANCIES AT RISK

- HYSTEROSCOPY (*it excludes uterine malformations*)
- MICROBIOLOGICAL CERVIX AND VAGINAL EXAM (*research for trichomonas; Candida! herpes; chlamydia.' mycoplasm*)
- CARPENTER TEST and OGTT (*to reveal an hidden or potential diabetes*)
- HORMONAL DOSAGES (*fertility control*)
- PELVIC ECHOGRAPHY
- ANTINUCLEUS ANTIBODIES RESEARCH, ANTI-DNA, ANTICARDIOLIPINA, LUPUS ANTICOAGULANT (*responsible for poll-miscarriage*)
- ARTERIAL PRESSION MONITORING IN THE 24 HOURS
- COUPLE SEROLOGICAL TEST (HIV, TORCH, HCV)
- TYPIFICATION OF THE ANTIGENIC OF HISTOCOMPATIBILITY ANTIGENS (HLA-DR) (*repeated miscarriages, pre-term labours, pre-clampsia*)
- SEMINAL LIQUID TEST (*to evaluate the fertility and any possible sperm abnormal condition*)
- DIAGNOSTIC, CYTOGENETIC AND MOLECULAR DNA INVESTIGATION (*in pregnant women subject to the risk of genetic diseases or diseases causing malformations*).

The prevention can be done:

- through preconception screening programmes, to identify couples at risk for few of the most common diseases, such as thalassemia.
- avoiding environmental factors, toxic substances, medicines, which are notorious to be mutable or cause of anomalies of the embryonic development.

*Anamnesis' Personal
Reproductive
Familiar
Environmental*

*Exams: Objective
Laboratory
Instrumental
Consultation*

