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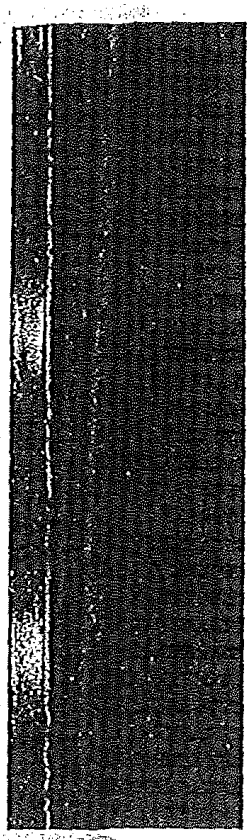

GENETIC COUNSELING

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ABSTRACTS
5th EUROPEAN MEETING ON PSYCHOSOCIAL ASPECTS OF GENETICS CAMPIDOGLIO, SALA DELLA PROTOMOTECA
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INNOVATIVE COUNSELLING FOR THE PREDICTIVE TESTING PROGRAMME FOR HUNTINGTON'S DISEASE

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In the light of five years experience and the disclosure of well over 200 results, we believe we offer a unique programme that is fully flexible to the individual needs of the consultands whilst, at the same time, respecting the International guidelines which are laid down for Predictive Testing for Huntington's Disease.

This paper will explore, with case examples, elements of the programme that we believe have contributed to its success. «Success» being measured by stated consultand well being following disclosure. The elements include the basic outline of the programme which may be extended, or in some cases contracted, depending on individual circumstances.

We emphasise the personal nature of the sessions with a single counsellor which has often been cathartic and healing for the consultand. This compares to some programmes in other centres where the consultand is seen by two or more medical personnel each time, which may be intimidating and block communication at a deeper level. A useful aid is the use of a personal diary or «Journal of Feelings» – this being kept for a 2 week period by the counsultand and, if applicable, their partner. One week imagining their result is normal and the other week that the result is that they have inherited the affected Huntington's disease gene.

This process has produced insights for many and gives the focus to the sessions.

Finally we have implemented a post test medical programme as an option for those who have inherited the expanded gene. This involves full neurological testing, clinical examination and a CAT-scan. This is offered every 1-2 years, or more frequently, at the request of the consultand.

As more inherited adult onset diseases are now DNA testable, the above model is adapted and is being applied at the Murdoch Institute.

PREDICTION OF PSYCHOLOGICAL FUNCTIONING ONE YEAR AFTER THE PREDICTIVE TEST FOR HUNTINGTON'S DISEASE

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For individuals at risk for Huntington's disease, the anxiety and uncertainty about the future may be very burdensome. For some at risk persons, this situation is at the origin of a request for predictive DNA-testing. The aim of this paper is two-fold. First, we want to evaluate whether knowing one's carrier on psychological functioning one year after the test. Second, we endeavor to identify pretest predictors of psychological adaptation one year after the predictive test (general anxiety, depression level and ego strength).

The impact of the predictive test result was assessed in 53 tested individuals, using pre- and posttest psychometric measurement.

provide training for non medical genetic counsellors to work alongside medical geneticists in regional genetic services. The course runs over 2 years and includes *taught modules, practical experience,* and a *research project* on a psychosocial topic. A maximum of 8 students are accepted each year from up to 100 applicants with backgrounds in genetics, nursing or related fields. *Taught modules* include human and clinical genetics, ethics, statistics and research skills, and a series of counselling modules. *Practical experience* includes first year community placements and second year placements in the genetic clinics. Each student completes a research project culminating in a dissertation and students are encouraged to publish and present their results at meeting such as this one. As of October 1996, 16 students have completed the course and all are employed in genetic counselling/research; 14 further students are in their first or second year of the course. Feedback has been favourable from students and employers, and several centres in the UK have plans to start up similar training courses.

The role of genetic nurses and counsellors in the UK has a much longer history than that of the Manchester MSc course. There is an Association of Genetic Nurses and Counsellors which is a constituent group to the British Society of Human Genetics. There are currently 100 members, and the Association recently completed a working party report on the role and practice of genetic nurses, headed by Heather Skirton. The results of a questionnaire sent to nurses and counsellors, as well as to medical geneticists, confirmed the broad and developing role of non medical practitioners in the UK. Genetic nurses and counsellors are involved in a team approach to genetic referrals, provide genetic counselling independently for more straightforward referrals, offer psychotherapeutic counselling, and contribute to teaching, research and administration.

ORGANIZATION OF G.C. IN ITALY AND INPUT OF PSYCHOSOCIAL DISCIPLINES: A PRELIMINARY STUDY

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In recent years, thanks to their new diagnostic potential, recombinant DNA technologies have revolutionized the approach to genetic diseases, opening up new possibilities for identifying and making a diagnosis of a growing number of hereditary disorders.

In Italy as well this has brought about an increase in the number of genetic consulting services and of laboratories where specific diagnostic procedures can be carried out.

In order to gain an understanding of the way in which these services are structured and organized we sent out a questionnaire specifically designed for this survey to 193 public and private centres listed in the Guide to the Diagnostic and Treatment Services for Genetic Disorders, which is the most updated list available in Italy at the moment.

The questionnaire consists of two parts:

- Part A, to be filled in by the person in charge of the diagnostic service.
- Part B, to be filled in by the person in charge of the service which deals with the psychosocial aspects of genetic counseling.

So far some 25% of all the centres to which they were sent have responded. However, the questionnaires duly filled in come from 13 out the 19 italian Regions which means that most of the national territory has been covered.

Some interesting remarks can be made on these preliminary data.

Data from Questionnaire A, concerning the organization of the centres, show that 98% of the centres that have answered carry out both diagnostic and counseling activities.

54% of the respondents states that the psychosocial aspects of g.c. are dealt with by doctors or biologists whereas 46% assigns the task of managing the psychosocial problems to social workers or psychologists.

82% deems that the training of the medical staff is insufficient to deal with such issues as: anxiety and feelings of guilt, difficulties in making reproductive choices and complex family and couple dynamics which are also indicated as being the most frequently encountered problems.

The psychosocial operators who answered to Questionnaire B are to be referred to 20 centres. Most of the respondents have begun to work in the area of g.c. only during the last 5 years. Hence this professional profile is a rather recent one in Italy.

42% of these operators have taken specific training courses on g.c. and all but one feel that it is indispensable.

They complain the lack of coordination with the doctors, the directiveness of the medical team, the insufficient exploration of the psychosocial problems and the lack of boundaries in professional accountability.

On the basis of these preliminary data, we can infer a fairly realistic picture of what is going on in Italy in this area, especially for the Northern and Central Regions of the Country.

The picture is one of a severe delay with regard to the care for psychosocial aspects of g.c. and in providing specific training courses for ad hoc operators.

The research is still on.

GENETIC COUNSELING IN THE COMMUNITY: A TEN YEAR EXPERIENCE IN NORTHEAST ITALY

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Genetic disorders produce a major burden on families and on society because of their frequency, morbility and mortality. The molecular revolution has led to preclinical diagnosis of an increasing number of genetic diseases, and to a better understanding of molecular pathology.

Genetic counseling (GC) is a complex medical process which provides affected individuals and their families with medical and genetic information. Several topics have to be covered: analysis of family members' requests, diagnostic procedures, natural history, occurrence/recurrence risk, and primary, secondary or tertiary prevention.

For these reasons there are two conflicting needs in the provision of GC, to centralize it, or to diffuse it into the community.

To achieve community-based genetic services two approaches could be evolved: a center-satellite system (CSS) with a center directly involved in genetic counseling using several community based screening centers, or first call genetic services where genetic counseling is provided by non-geneticist with the center involved only in complicated situations. The ideal model might use both.

A 2 level system in North East Italy was organized. This system worked from the early '80s to the early '90s. The CSS adopted foresaw a regional genetic service acting at 2nd-level, and satellite clinics in the community hospitals of this area, acting autonomously for first-call counsultations, and in the meantime collecting referrals for more complicated situations and contributing to 2nd-level consultation with the genetic center staff.

If the referred case was considered to be a 1st-level consultation, the medical evaluation of the proband, the analysis of available data, the request for further information, the collection of the new data by the genetic associate (GA), the reanalysis of the situation, the genetic counseling, and the follow-up to check understanding of genetic information were locally done.

If the referred case was considered to be a 2nd-level consultation the collected data were transmitted to the center, where they were analyzed. A medical evaluation of the proband on the site of the satellite clinic was subsequently performed by the staff of the center together with the satellite clinic staff. Locally the organization of requests (information, tests) was carried out. The data were re evaluated by the staff of the center, and finally a genetic counseling session was performed at the satellite clinic. The clinical follow-up was carried out locally by the MD and the follow-up for the consultants' understanding by the GA.

The number of genetic consultations by level grew over time. The 1st-level consultations greatly increased over time (10 in 1981, 361 in 1990) overtaking, as expected, 2nd-level ones (99 in 1981, 260 in 1990). The total number of consultations reached 600 per years.

Four years after the termination of this research project, 5 satellite clinics are no longer working, while the other 6 are still active and fully autonomous either for 1st or 2nd-level consultations.

The only links with the center are a clinical genetics meeting every 3 months, dedicated to the presentation of dysmorfic cases referred for genetic counseling, and occasional training session for MDs in the university center.

TEACHING A COUNSELLING APPROACH FOR GENETIC COUNSELLING PRACTICE

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An approach to teaching counselling skills to clinical geneticists and non-medical co-workers has evolved from our experience of running courses for both groups.

We offer a two year Masters degree course (MSc) in genetic counselling aimed at nursing and other graduates, and a short residential workshop for doctors training in clinical genetics. The demand for both courses has been high and the feedback from participants has been very positive.

Despite the wide range of backgrounds and counselling experience among both clinical geneticists and MSc genetic counselling students, it has been helpful to begin with more «basic» skills including listening skills, empathic responses, question style and recognition of verbal and non-verbal cues. These skills are demonstrated in videotapes of structured interviews aimed at eliciting patients' concerns. This provides a basis for further sessions addressing strategies to deal with more difficult genetic counselling situations such

Female consultees who were first degree relatives of cancer patients and who had at least one case of breast cancer in their family participated in this study.

Among the 248 eligible consultees attending the clinics during the study period, 84.3% answered a post-consultation questionnaire. Among the 209 respondents, 40.7% (n=85) were cancer patients and 59.3% (n=124) were healthy consultees.

A high consensus in favor of genetic testing was noted, since 87.7% of the sample stated that they would ask for breast cancer gene testing if this test became available.

The underlying assumption of 96.6% of the women was that their health surveillance would be improved after a positive test. High awareness of the anxiety that would be generated in a family after positive results was observed and found to be associated ($p<0.05$) with the anxiety and depressive profiles of the patients.

Half of the healthy respondents said they would not change their attitude towards screening if the results of predictive testing turned out to be negative. Only 13.7% of the 161 patients who stated that the oncogeneticists asked them to contact their relatives firmly refused to do so, mainly because of difficult family relationships.

PREDICTIVE TESTING FOR FAP - HAS THE HUNTINGTON'S DISEASE PREDICTIVE TESTING MODEL BEEN A HELP OR A HINDRANCE?

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It has been proposed that the Huntington's Disease Predictive Testing model may be an appropriate one for families undertaking predictive testing for familial cancer syndromes.

Predictive testing for FAP has been available at the VCGS for over twelve months. Using the Huntington's Disease model as a guideline, we have introduced a three session program for individuals requesting predictive testing. Session one, where the family is seen as a group by the medical geneticist and the genetic counsellor, is where the pedigree and the testing process are explained to the family.

Session two is where family members are seen on an individual basis to explore the ramifications of taking the test, as an individual and as a member of a family.

Session three is where the results are disclosed.

The outline of these sessions is a basis and not a rigid plan and the sessions may be changed depending on the individuals needs and wishes.

This presentation will examine in what ways the Huntington's Disease model has been helpful in our testing program for FAP, in what ways it has been a hindrance and what we can learn from the HD model to take with us, as counsellors, for the next era of those individuals seeking predictive testing for familial cancer syndromes.

COLOURED PROGRESSIVE MATRICES: ERROR TYPE IN DEMENTIA AND MEMORY DYSFUNCTION

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The importance of the cognitive assessment in determining early diagnosis of neurological diseases, is widely emphasized. Nevertheless, not much has been made for the development of adequate tools of investigation.

These tools must be easy to administer and with a high sensitivity and specificity for different disorders.

This work describes the results obtained with Raven's Coloured Progressive Matrices (CPM), a test designed to assess the intellectual processes of children, mentally defective individuals and eldery people. The test was administered to 92 subjects (mean age= 66.2, sd=11.3) belonging to 3 different groups: 31 demented patients, 34 dismnesic patients and 27 normal subjects. All subjects underwent a comprehensive neuropsichological examination that included, among other tests, the Mini Mental State Examination (MMSE).

CPM scores resulted to be different across groups: demented patients had (CPM=12.3) lower scores than dismnesic (CPM=22.3) and normal (CPM=27.4) subjects. The same findings were obtained with MMSE scores.

Considering CPM<20 and MMSE<26 as cut-off scores for demented patients, we obtained 90% of sensitivity.

When dismnesic patients are considered, the MMSE categorizes as inferior 53% of the patients, while the CPM identifies as such only the 35%.

When CPM errors are analysed, instead of the correct responses, the best diagnostic utility of this test results evident.

A factorial analysis conducted on the different types of error reveals 2 factors: a factor choice and a factor orientation. On the first type of error,

dismnesic patients perform like normal subjects, while on the second type of error demented and dismnesic present an equal number of errors. This result is very important to diagnostic goals, since the first type of error might be more closely related to a diffused degeneration, while the second type might be caused by alterations of specific cerebral structures.

DEVELOPMENTAL DYSLEXIA AGGREGATING IN ITALIAN FAMILIES

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Developmental Dyslexia (D.D.) is a deficit both in learning and in mastering reading, in spite of average intelligence, adequate educational opportunities, and in lack of neurological and sensorial deficits. Troubles in writing and calculations are often associated, too.

At present probably many researchers agree about the deficit definition, but not about its aetiology. There is a variety of causes and neuropsycological profiles of D.D.. However, many studies describe D.D. as aggregating in families, and in some cases the genetic base of this deficit has been evidenced (Finucci *et al.*, 76; De Fries *et al.*, 93).

The aim of the present study is to start a research on aggregating of Developmental Dyslexia in Italian Families.

64 Italian subjects with D.D. have been assessed in 1994 and 1995 at the Regional Center for Linguistic-Cognitive Deficits in Bologna and at the Clinical Psycology-Neuropsychology Service of «Bambino Gesù» Pediatric Hospital in Rome. Data from direct interviews with the relatives of all the subjects and from the neuropsychological assessment of 15 probands, their parents and, in some cases, their brothers and sisters, will be reported.

AN INTEGRATED MODEL OF PSYCHOLOGICAL DIAGNOSIS OF CHILDREN WITH GENETIC SYNDROMES AND THEIR PARENTS

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Among the causes of mental disfunction, the genetic syndromes assume greater importance in both a quantitative sense (Kallen, 1988) and a qualitative sense (psychological profiles in mental retardation, Masi, Stella, 1995) The age at diagnosis is high and so children and parents face extended diagnostic procedure with risk of physical weakening and mental break-down.

For more than ten years, in our hospital, the units of clinical psychology- neuropsychology and genetic medicine have used an integrated diagnostic procedure for their patients, taking into account the different ways of access often used.

Our system of diagnosis envisages simultaneous observations of children and parents during a parallel course conducted separately by different specialists in adjacent rooms.

During the initial phase the personal history of the child is confused with that of the parents.

In the final phase, through a process of elaboration, the behaviour and emotions, the ability and competence of the children are individualised and separated from those of the parents.

Through an integrated comprethension of the biological and psychological aspects, one tries via periodic checkes to reconstruct the sense of self and the identity of these children which are sometime fragmentary as well as the educational competence of the parents often «out of order»

STUDY OF THE FACTORS THAT INFLUENCE EATING ATTITUDE IN CYSTIC FIBROSIS PATIENTS

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Psychosocial factors may play an important role in CF malnutrition. In our study, 67 CF patients (31 females, 36 males, aged 10 to 32 years) were evaluated by Ch-EAT questionnaire. Average Ch-EAT score in the series was 7.6; the average score in females was higher than the average score of males (9.5 and 5.9, respectively). In our series, 5 patients (3 females, 2 males) had a Ch-EAT score higher than 20, ranking in the anorexia nervosa range; another 8 patients (6 females, 2 males) had a Ch-EAT score between 15 and 20, definible as borderline. These 13 patients were