

Outcome of genetic counseling: the experience of a single centre in Italy

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DEFINITION

Genetic counseling (GC) is a nondirective communication process that addresses the individual's and/or the family's need, regarding a hereditary disorder, to:

- ◆ understand the diagnosis (pre- or postnatal), the disease's course, management and possible treatment;
- ◆ understand the genetic basis of the disease and the recurrence risk;
- ◆ understand the available options to cope with disease risk;
- ◆ understand the reproductive options;
- ◆ face the most appropriate choice, taking into consideration the disease risk and the family's principles and ethics;
- ◆ accomplish the best possible adaptation to the disease.

All these views of GC encompass considerable psychological and ethical merits. The counselor should be able to permit consultant's full decisional autonomy. Recent progress in genetics, including the acquisition of new knowledge, the diffusion of genetic testing and the make-up of new professional figures, has brought to a rapid growth of the demand for GC. As a consequence, an evaluation of the outcome of GC is essential, in order to determine the quality and optimize the service,

MATERIALS & METHODS

This study has been based on total data on GC sessions held in the C.S.S.-Mendel Institute in Rome, by the same clinical geneticist, during a period of 30 months (July 2004-December 2006). Data has been collected through Shire®, a specific GC management software. The impact of prenatal GC has been evaluated retrospectively through the administration of a multiple choice questionnaire in anonymous form, to 252 couples, a few months after delivery. The questionnaire included a first part about pregnancy evolution and a second one, about the GC received.

The Mendel Institute in Rome



Clinical activity	m ²	178
Laboratories	m ²	1,293
General services	m ²	895
Total		m² 2,166

RESULTS & CONCLUSIONS

Type of new information provided / GC sessions concluded with a final report (%)

1. Diagnosis	471/898 (52%)
2. Occurrence/recurrence risk of a disease	463/898 (51,5%)
3. Etiology/pathogenesis of the disease	282/898 (31%)
4. Indication for invasive or non-invasive prenatal diagnosis	223/898 (25%)
5. Interpretation of a prenatal diagnosis result (invasive and non-invasive)	191/898 (21%)
6. Disease treatment/management	186/898 (21%)
7. Indication for a genetic test	132/898 (15%)
8. Genetic test availability	120/898 (13%)
9. Prenatal diagnosis (invasive and non-invasive) availability	109/898 (12%)
10. Clinical implications of the disease	88/898 (10%)
11. Request of in-depth instrumental investigation	74/898 (8%)
12. Request of an external specialist consult	30/898 (3%)

◆ A final report, written in 79% of cases, is useful for the patient to acquire and understand information provided that should be accurate and accessible to other specialists.

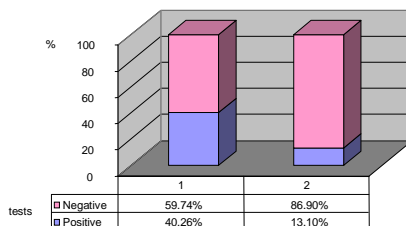
◆ In-depth investigation and collaboration with other specialists was necessary in 7% of cases while in 17% of cases samples for genetic tests were sent abroad, as most genetic diseases are rare and multisystemic.

◆ In 35% of GC postnatal sessions a diagnosis has not been accomplished, parallel to literature data. These were mostly cases of children with dysmorphism and encephalopathy.

◆ A genetic test has been requested in 25% of GC sessions. GC remains a principally medical act despite the progress in molecular genetics. Laboratory diagnosis is secondary to a well-founded clinical suspicion. Tests are not available for many genetic diseases and patients have frequently already performed tests in absence of clear indications or secondary to a false clinical diagnosis.

◆ The difference of positive molecular and cytogenetic tests results could be justified on the basis of specific clinical suspicion for molecular ones while cytogenetic tests are usually performed in the presence of non specific clinical presentation (psychomotor retardation associated or not to dysmorphism or multiple congenital malformations).

Results of genetic tests requested, molecular (1) and cytogenetic (2).



Regional provenience of patients referred



◆ The increased migration of patients from the South of Italy could be secondary to the absence of a clinical genetics center in their region, long waiting lines, the search for an expert or of a centre of reference.

◆ Fifty-nine % of couples referred for prenatal GC returned the filled-in questionnaire. Eighty-six % of them have been completely or considerably influenced by GC in their decision-making, witnessing the critical role of the counselor.

◆ The percentage of pregnancies at term was lower in cases of high risk of affected fetus ($\geq 15\%$), but it was not related to counseling efficacy as measured according to the consultants' perception. However the low number of cases and the variable social-cultural backgrounds of couples could represent a bias of ascertainment. These data underline the non-directiveness of GC.

◆ Aged mothers (over 35 years) with no children had less propensity to terminate pregnancies in respect to those with one or more children. According to our study, the decision of a couple to continue or not a pregnancy, in particular, in case of high risk of affected fetus, associates with maternal age and previous family history.

Counseling's burden on parents' decision, based on interview.

	SAME AS POPULATION RISK	1-3% ADDITIONAL RISK	HIGH ADDITIONAL RISK ($\geq 15\%$)	ONLY INFERTILITY RISK	TOTAL
Couples completely influenced	54/55 (98.18%)	10/10 (100%)	3/11 (27.27%)	9/9 (100%)	76/85 (89.41%)
Couples considerably influenced	22/24 (91.66%)	6/6 (100%)	1/6 (16.66%)	3/3 (100%)	32/39 (82.05%)
Couples little influenced	5/5 (100%)	—	—	2/2 (100%)	7/7 (100%)
Couples minimally influenced	5/5 (100%)	2/2 (100%)	3/5 (60%)	—	10/12 (83.33%)
Total	86/89 (96.62%)	18/18 (100%)	7/22 (31.81%)	14/14 (100%)	125/143 (87.41%)

At term pregnancies based on risk, previous family history and maternal age.

	SAME AS POPULATION RISK	1-3% ADDITIONAL RISK	HIGH ADDITIONAL RISK ($\geq 15\%$)	INFERTILITY RISK	TOTAL
Women ≥ 35 years old without children	9/9 (100%)	9/9 (100%)	2/3 (66%)	11/12 (92%)	31/33 (93.93%)
Women < 35 years old without children	21/21 (100%)	9/9 (100%)	4/12 (33%)	4/4 (100%)	38/46 (82.60%)
Women < 35 years old, with 1 or more children	4/5 (80%)	3/3 (100%)	1/4 (25%)	2/2 (100%)	10/14 (71.42%)
Women ≥ 35 years old, with 1 or more children	21/21 (100%)	8/8 (100%)	0/5 (0%)	8/8 (100%)	37/42 (88.09%)
Total	55/56 (98%)	29/29 (100%)	7/24 (29%)	25/26 (96%)	116/135 (85.92%)