Cancer genetic counselling

Counselling in cancer genetics may differ from genetic counselling in cancer.

Cancer risk counsellors must provide patient education, risk assessment, pre- and post-test counselling, and intervention planning. In this complex multistep process, Contegiacomo et al. [1] favour the central role of the oncologist. Yet in many countries, including Italy, cancer genetic counselling is performed by trained clinical geneticists [2, 3]. It is our opinion that no matter who is the cancer genetic counsellor, some points must be addressed clearly.

In the proposed multistep model, pedigree construction is performed only as a second step (T1), after an extensive information/education session (T0). We believe that the inclusion in a genetic risk assessment model should be performed only after collection of an accurate personal and familial history, rather than as a second step, after extensive information and education to the patient, because ‘collecting genetic information is the first and most important step in genetic counselling’ [4].

Why should we extensively inform consultants about hereditary, familial and sporadic forms of cancer, available methods for risk assessment, implication of cancer risk, strategies for risk management, surveillance and prevention before knowing whether they have a cancer risk other than the normal population? General information about common risk factors for cancer development is provided through easy-to-read papers, pamphlets, educational websites and so on through Regional Health Services, while pedigree drawing is the way to collect relevant clinical history in a specific condition, and is the essential step with which to begin any genetic counselling.

Cancer genetic counselling is now offered not only in a research project framework, but also as a medical practice in several clinical settings where other inherited late-onset conditions, with similar or more critical psychological impact, are followed.

Finally, the statement that ‘a low cognitive level and psychological distress preclude continuation of counselling’ cannot be accepted in a genetic counselling setting. In our opinion, the patient’s psychological distress should not preclude the practice of genetic counselling. The impact of psychological distress in counselling was analysed by psychologists involved in presymptomatic testing of late-onset disorders and is regarded as a part of the process of decision-making. DudokdeWit et al. [5] showed that risk carriers of late-onset diseases with higher stress scores may be actively dealing with the emotional implications of the test, whereas those with low stress scores may be unable to face these implications. Therefore, it seems important to identify the patient’s strategy of coping with the potential threat, in order to provide suitable counselling and necessary guidance.

References

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Touch imprint cytologic preparations and the diagnosis of head and neck mass lesions

The diagnosis of head and neck mass lesions entails examination of initial frozen sections, followed by the evaluation of permanent histological sections. This process is worrying for the surgeon who is unsure about the tentative diagnosis of the frozen sections. Moreover, the pathologist should make a hurried diagnosis on suboptimally prepared specimens. To date, controversial reports are available about the use of touch imprint cytology (TIC) in the diagnosis of these lesions [1–3]. To assess the utility of an intraoperative TIC in the diagnosis of these lesions, 30 head and neck masses (nasal, pharyngeal, laryngeal and oral lesions) were examined by TIC, permanent histological sections and immunostaining methods. Immediately after obtaining the biopsy specimens, and prior to placing them in fixative, each specimen was imprinted on several glass slides (TIC), fixed immediately and stained with haematoxylin & eosin. The cytological results were reported as: (i) malignant (the cellular findings are diagnostic of malignancy); (ii) suspicious for malignancy (suggestion of cancer but uncertain due to limited number of cells or to degree of atypia); (iii) negative for malignancy (no evidence of malignancy); or (iv) unsatisfactory specimen (scant cellularity, air drying or distortion artifact, obscuring blood or inflammation) [4]. The cytological interpretation was carried out intraoperatively. His-
Histological diagnosis | Histological sections (biopsy specimens) | Touch imprint cytology
---|---|---
Positive for malignancy | 15 | 16
Carcinoma | 12 | 
Lymphoma | 3 | 
Sarcoma | 0 | 
Suspicious | | 2
Negative for malignancy | 15 | 12
Inflammatory lesions | 6 | 
Benign tumor | 2 | 
Non-neoplastic polyp | 3 | 
Goiter (colloid and toxic goiter) | 4 | 
Unsatisfactory | 0 | 
Total | 30 | 30

Table 1. Correlation between the results of the biopsy specimens and touch imprint preparations in patients with head and neck mass lesions.

The cytological examination of the permanent sections was carried several days later.

The cytological evaluation of the TIC revealed 12, 16 and two cases as benign, malignant and suspicious for malignancy, respectively. Further histological examination of the permanent sections revealed 15 cases each as malignant and benign lesions. The concordance between touch imprint and paraffin sections was 90% (27 of 30). The sensitivity and specificity of TIC in detecting malignancy were 88% and 92%, respectively (Table 1). These high rates highlight the value of TIC as a relatively simple technique that can allow the pathologist to render an intraoperative diagnosis. In addition, TIC is less expensive than the frozen section method. The disadvantages of the TIC method are that it does not provide architectural information and it cannot distinguish between in situ and invasive lesions. The cytological diagnosis ‘suspicious for cancer’ was encountered in two cases. In these two cases, cellularity was lacking, and many bare nuclei and benign cell clusters were present. The cases suspicious for malignancy were negative on further permanent histological sections. We propose the necessity of maintaining this diagnostic category for two reasons. First, it allows the cytologist to raise the suspicion of malignancy in a mass lesion that does not meet all the TIC criteria for malignancy. Secondly, this diagnostic category helps keep false-positive diagnoses near zero. The false-negative cases in our series (two cases) may be inherent in the procedure. With further analysis of these cases, the missed diagnosis could have been averted by careful screening to detect scant malignant cells. In frozen sections, many factors contribute to the false-negative rate, including the suboptimal preparation of the specimen and sampling errors [5]. Our results indicate that TIC can overcome its deficits, and proved useful in evaluating head and neck mass lesions (nasal, pharyngeal, laryngeal and oral lesions).

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Retroperitoneal lymph node dissection versus chemotherapy for stage I testicular nonseminomas

The European Germ Cell Cancer Consensus Group’s statement on the diagnosis and treatment of germ cell cancer provides an excellent review of our current state of knowledge and is a welcome addition to the literature [1]. Nonetheless, I am surprised by the omission of any acknowledgement of the significant cardiovascular morbidity associated with platinum-based chemotherapy in men with germ cell tumors. This is particularly relevant with regard to treatment options for men with clinical stage I nonseminomatous germ cell tumors. When men are deciding between retroperitoneal lymph node dissection (RPLND) and primary chemotherapy, it is essential for them to be educated about the potential for serious long-term toxicity from cisplatin-based chemotherapy. Huddart et al. [2] conducted a study of cardiovascular events in 992 men with a history of testicular cancer and reported that exposure to platinum-based chemotherapy was associated with a relative risk of cardiovascular events of 2.59 [95% confidence interval (CI) 1.15–5.84] compared with men treated...