EP14. Professional responses to nondisclosure of genetic information

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The patient's right to privacy, and the professional's obligation to respect their confidentiality, are both challenged by the argument that genetic information is shared, belonging to a family rather than the individual. If important genetic information is not passed on, then relatives could sometimes be at avoidable risk of disease complications. Professionals are concerned that they may be legally held liable when such complications arise, and are beginning to alter the wording of consent forms for genetic testing to coerce those being tested to make their results available to others.
The arguments used to justify force disclosure or coerced consent to disclosure often fail to take account of the temporal dimension or the medical uncertainty about the potential for avoidable harm form non-disclosure. We present three cancer genetic counselling cases where non disclosure was recognised as an issue, but supportive contact was maintained with the client and - after some years- they informed their relatives about their genetic status. The benefits to the clients and their family relationships from allowing them time for such decisions are very substantial and are easily omitted form bioethical discussions of the potential harm of non-disclosure.

EP15. Facilitating family communication about predictive genetic testing for HNPCC: the proband's perception.

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Although it is known that genetic information is rarely completely shared in families, genetic professionals rely on family members to notify relatives that predictive testing for cancer predisposition is available. This task usually falls to the proband - the affected individual in whom the causative mutation is first found. Probands can find this a burden and professional support during this process has been recommended (Bonadona et al, Cancer Epidemiol Biomarkers Prev 11: 97-104).
With the goal of enhancing support for probands and maximising information dissemination, we explored the utility of genetic counselling and communication aids to probands informing relatives that predictive testing was available for Hereditary Non Polyposis Colorectal Cancer (HNPCC).
Five men and seven women were interviewed by phone.
Respondents reported few difficulties in communicating information about testing. Nonetheless, all respondents failed to inform some biologically, socially or geographically distant relatives. Generally, respondents would not change the way they informed family members, although it appears that men may require more guidance and support during this process. Probands conceded that aids such as letters would be helpful for *other* people who had communication problems, but were seen as a source of accurate information rather than a way of informing additional relatives.
Respondents found communication relatively easy and probably represent the 'best case' scenario. Genetic services should consider a more active role, working in partnership with the proband to ensure that all at-risk family members have access to predictive genetic testing.

EP16. Exploring families' psychosocial needs after genetic diagnosis: Perceptions of genetic counselors.

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A new genetic diagnosis is likely to raise significant psychosocial reactions in a family. There is a body of literature on the psychological impacts of these diagnoses, but only anecdotal evidence about other psychosocial disruptions. In addition, because, genetic counselors in the United States do not usually have long-term contact with their clients, there is little know about the long-term psychosocial needs of families with a new diagnosis. I report here on the first of two pilot studies of families' psychosocial needs. The first study focuses on what genetic counselors understand about families short-term needs. The second study will be based on a patient and family sample and will expressly include long-term adjustments.
We explored the knowledge and practices regarding psychosocial issues with the primary provider of genetic services, genetic counselors. We interviewed 20 genetic counselors about the psychosocial issues dealt with in counseling sessions, what services exist, and client barriers to using the services. Genetic counselors identified a complex set of needs for psychological, support and concrete services. Most counselors address likely anticipate and discuss psychological responses and family social support with patients. Few discuss such other psychosocial issues as coping strategies, environmental resources, and changes in family roles. Counselors discussed barriers to comprehensive psychosocial services, and most often cited their lack of training, limited time with clients, and external barriers, most importantly a lack of insurance coverage for care. Counselors acknowledge that many families need long-term follow-up care, and often refer patients to social workers for those services.

EP17. Rehabilitation treatment in HD: benefits beyond motor and verbal improvement.

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Huntington's disease (HD) is a neuro-degenerative, autosomal dominant, late-onset disease characterized by slowly progressive movement disorders, cognitive deterioration and psychiatric manifestations.
In 1999 we started a rehabilitation protocol for HD patients at the Caring Home «Nova Salus» of Trasacco, in Abruzzo Region. The interdisciplinary treatments were performed at an intensive regimen for 8 hours a day per 6 ½ days (Saturday only in the morning, Sunday free), for 3 weeks for a maximum number of 3 admissions per year.
The effect of the rehabilitation was evaluated both in terms of motor performance, quantitatively assessed through motor scales, and in terms of subjective evaluation by patients and caregivers. Here we report the results obtained through the latter approach.
An ad hoc Questionnaire was devised and sent to 59 subjects. Forty-five of them (76%) sent back the filled in questionnaire.
Overall positive effects of the rehabilitation experience were reported by 97.6% of respondents. Improvements were reported for body control (89.7%), speech (85.3%), balance (81.3%), gait (80.9%) and swallowing (80.9%). Positive effects were also reported for several psychosocial aspects, namely mood state (89.4%), establishing new friendships (88.8%), reducing apathy (81,3%), family relationship (78%) and social relationship (74.4%).
The positive, although temporary, effects obtained with the treatment will constitute the base for developing education programs for health care providers in our country.

EP18. Diagnosis of a non-life-threatening, progressive, neuromuscular condition. ‚The impact on life'.

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Receiving a diagnosis for a progressive, neuromuscular condition such as Charcot-Marie-Tooth disease (CMT) or Myotonic muscular dystrophy can be news of major gravity. Initial symptoms of such conditions are often rationalised through activities of daily life, accepted as 'personal traits' or simply misdiagnosed for many years. A correct diagnosis is often not obtained until well into adulthood, as symptoms become more pronounced with age. This revelation can have numerous implications for an individual and their whole family. However literature on impact of diagnosis in progressive, non-life-threatening conditions is scarce.
This study collated data from 25 questionnaires. A series of open questions explored the impact on life following diagnosis of CMT or myotonic dystrophy. An 'Impact of Event' scale provided a quantified measure of the degree of diagnostic impact. 6 in-depth interviews gave greater insight into personal experiences of diagnosis.
As initial shock subsides post-diagnosis, a need for more information

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