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79 – From Familygram to Personalised Medical Care

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Familygram

As a profession, family medicine has a myriad of specific tools designed for assessing every single patient in the context of his/her family and its psycho-sociological and health circumstances. Even though many modern techniques have been introduced into clinical care, family physicians continue to use tools deeply rooted in simple family assessment techniques, considering them a unique privilege of the profession. Patient/family assessment is provided by the Family Apgar, Family profile and Family history. Through analysis, a family tree and family genogram can be established. Connection and assessment is made of all obtained data results in a familygram profile, which is recognised as an integral presentation of the entire family and its social and health record, including personal comments.

Family Apgar is designed to test five areas of family function (**A**daptability, **P**artnership, **G**rowth, **A**ffection, **R**esolve). Every question is scored as 2 (almost always), 1 (sometimes) and 0 (hardly ever). A total score of 7-10 presents a highly functional family and 0-3 a severely dysfunctional family.

Family profile comprises of different assessments. Family structure and family form show a large variety of different, nationally or regionally specific patterns, often strongly connected to different cultural backgrounds or family policy models. Some new types of families represent different environments that affect family members and their attitude to social and health-related behaviour.

Family history (FH) has become evident as the gateway to medical genetics (recognizing inherited disorders). Creating a FH involves several steps, the family medical history questionnaire often being the first step in this process. Patients may be encouraged to contact relatives to obtain or confirm information. The face-to-face interview is a critical part of eliciting family history, with the ultimate goal to obtain a pedigree that includes at least 3 generations (age or year of birth, age and cause of death, ethnic background of each grandparent, relevant health information -e.g., height and weight-, illnesses and age at diagnosis, information regarding prior genetic testing and information regarding pregnancies).

Family history plays an important role in identifying patients who may benefit from predictive genetic testing. A positive FH has been recognized as a risk factor for the development of cardiovascular diseases (CVD), diabetes (DM) and various cancers, moreover, it has been identified as an independent risk factor for the development of CVD and DM. It is known that family history of common chronic disease is associated with a 2- to 5-fold relative risk of developing the condition, and this increases with the number of affected relatives.

IT supporting systems (ITSS)

Several IT supporting systems (ITSS) have been developed to integrate all elements of familygram. Some of them are oriented to pedigree profiling and offer a standardized system for pedigree storage. Other pedigree oriented ITSS are particularly intended for epidemiologists in the sense that they allow customized automatic drawing of large numbers of pedigrees and remote and distributed consultation of pedigrees. Some other pedigree ITSS fully integrate risk analysis and support for industry standard databases and are designed for genetic counsellors and work with genetic marker data, enable haplotyping and allow exports to a range of linking analysis packages.

Family tree programmes enable construction of family trees, offering an ability to import data from other genealogy programmes. Attractive charts and reports in a variety of formats can be drawn and the photos, audio, and video files can be added to the tree.

Graphical genotyping package ITSS combines genetic map data and locus data to display graphical genotypes. Data can be viewed by individual or by linkage group, and sorted on markers. Statistics are calculated, and simultaneous filtering/selection on multiple linkage groups are made possible.

However, gathering, filtering and inputting data is always a doctor's task, while ITSS only manages inputted data. If the inputted data are more complete, the output and suggested course is more comprehensive.

Future challenge: a personalized medicine

In the future, medicine will become more and more focused on personalized approach to the patient. Personalized health care (PHC) is a strategic approach to medicine that is individualized, predictive, preventive, and is based on an individual's specific profile. People subject to increased risk because of their family history are among the most appropriate candidates for genetic profiling to find possible new biomarkers. Biomarkers can help in better understanding of the disease, prediction of its onset and development. Such genomic approach can help to evaluate co-morbidity of selected chronic diseases horizontally and connect them with family history. This shall help to reveal new horizons of genomics in chronic diseases.

Such processes should be supported by an IT centralised vendor-neutral clinical data repository with support for real-time transactional health data storage, query, retrieval and exchange. This is structured data set for the validation of the decision support model, efficiency calculations and bio-informatic studies.

Views on personalized medicine can vary according to standpoint: for patients, it offers access to life or better quality of life; researchers see potential in their own areas of specialization; for pharmaceutical industry it offers the hope of developing more effective medicines as understanding of specific diseases advances; a physician sees a new way of treating a patient; for society, it may be perceived as offering scope for an overall improvement in healthcare – or a new cost.

It will be possible for patients and professionals to discuss options only if a common language and conceptual understanding of genetic and protein signatures and pathophysiology of the disease are available for both – taking account of the individual patient's psychological make-up (educational level, mental state, and attitudes to risk). Additionally, ethical standards must be evaluated taking into account the specifics of PM and creating an infra-structure in which genetic information is available in a regulated context.

To conclude, familygram opens the pathway to PM which is entering medical care. However no one, not even ITSS, can replace the family physician and its fundamental role in assessing the health of each individual within the context of family, society and socio-economic aspects and to communicate with the patient on his/her personal level.

Take home messages

- Familygram is an important tool for assessing every patient in the content of his/her family.
- Family history enables detection of risk factors and presents a gateway to medical genetics.
- IT supporting systems (ITSS) have been developed to integrate all elements of familygram.
- Medicine is becoming focused on a personalized approach to patient in the near future.
- Nobody can replace the family physician and its fundamental role in assessing the health of each individual.

Original abstract

http://www.woncaeurope.org/content/295-family-profile-and-familygram-99

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