Brief Report
Volume 12:7, 2021

Journal of Molecular Biomarkers & Diagnosis

ISSN: 2155-9929 Open Access

Population Genetic Screening Programmes

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Brief Report

This paper examines the professional and scientific views on the principles, techniques, practices, and policies that impact on the population screening programmes in Europe. This paper focuses on the problems surrounding potential screening programmes, which require further discussion before their introduction. It aims to extend, among the health-care professions and health policy-makers, awareness of the potential screening programmes as a problem of accelerating concern to public health. The methods comprised primarily the review of the prevailing professional guidelines, regulatory frameworks and other documents associated with population screening programmes in Europe. Then, the questions that require debate, in reference to differing types of screening before and after birth, were examined. Screening for conditions like CF, Duchenne dystrophy, hypercholesterolemia, fragile X syndrome, hemochromatosis, and cancer susceptibility was discussed. Special issues associated with screening were also examined, like consent, family aspects, commercialization, the players on the scene and monitoring screening programmes. It has been questioned whether screening differs from other sorts of screening and testing in terms of ethical issues. The overall impression on the longer term of screening is that one wants to 'proceed with caution', with more active impetus from the side of patients' organizations and more reluctance from the policy-makers. The latter attempt to obviate the potential problems about the abortion and eugenics issues which may be perceived as a greater problem than it's actually. However, it seems important to take care of a balance between a 'professional duty of care' and 'personal autonomy'.

Genetic screening refers to explicit and systematic programmes directed either at whole populations of asymptomatic individuals or at subpopulations during which risk is understood to be increased. Screening has got to be distinguished from genetic testing because the implications are different. Genetic testing is administered on patients who for whatever reason have taken the initiative and seek professional advice. In screening, tests could also be seen to be imposed on individuals. the moral dilemmas are magnified and therefore the responsibilities for the physician correspondingly greater.2 The genetic nature of a disorder leads to risk implications to relations of the person screened, albeit they'll not be, nor perhaps wish to be, included within the screening programme. Screening is additionally distinguished from other sorts of screening because it doesn't necessarily cause the prevention or treatment of diseases.

The methods used for analyzing the principles, the techniques, the practices, and therefore the policies of screening were primarily the review of the prevailing professional guidelines, regulatory frameworks and other documents associated with population screening programmes in Europe. Then, with the assistance of the prevailing guidelines and a review of literature, the tactic was to look at questions that require debate, with reference to differing types of screening before and after birth. Screening for conditions like CF (CF), Duchenne dystrophy (DMD), hypercholesterolemia, fragile X syndrome, hemochromatosis, and cancer susceptibility was discussed. Special issues associated with screening were also examined, like consent, family aspects, commercialization, the players on the scene and monitoring screening programmes. Afterwards, these questions were debated during a world workshop organized by the ecu Society of Human Genetics Public and Professional Policy Committee in Amsterdam, Netherlands.

The purpose of the workshop was to spot, from knowledgeable viewpoint, the foremost important/pressing/burning ethical issues concerning population screening programmes in Europe. the problems were discussed throughout the subsequent four sessions: (1) general aspects of genetic screening; (2) programmes before birth; (3) programmes after birth; and (4) ethical issues raised by such programmes.

General principles for screening

Both at the national and supranational levels, guidelines are elaborated with a view to the developments in screening and therefore the ethical issues raised by it. of these documents affect the question on which requirements apply to screening programmes. In any screening programme, guidelines should be established governing its aim, limitations, scope, and ethical aspects, also because the storage and registration of knowledge or material, the necessity for follow-up (including social consequences), and therefore the risk of side effects. The 2 most often cited objectives of screening are to scale back the prevalence of the disorder and to tell individuals and couples in danger about their reproductive choices. Particular attention is being paid to the rights of participants in terms of consent, confidentiality, and data protection.

- Genetic screening should be voluntary, not mandatory;
- Genetic screening should be preceded by adequate information about the purpose and possible outcomes of the screen or test and potential choices to be made;
- Anonymous screening for epidemiological purposes may be conducted after notification of the population to be screened;
- Results should not be disclosed to employers, insurers, schools, or others without the individual's consent, in order to avoid possible discrimination;
- In rare cases where disclosure may be in the best interests of the individual or of public safety, the health provider may work with the individual towards a decision by him/her;
- Test results should be followed by genetic counselling, particularly when they are unfavorable;
- If treatment or prevention exists or is available, this should be offered with a minimum of delay;
- New-borns screening should be mandatory and free of charge if early diagnosis and treatment will benefit the newborn.

Are population screening programmes needed? Genetic testing is now a well-established a part of medical aid, which can benefit variety of various groups of people. Given the rarity of great genetic conditions, the event of screening programs, even within the light of latest genetic knowledge, won't cause the testing of all individuals. Instead, the offer of a test to an outsized subgroup of them, with the aim of identifying those at higher risk in order that more specific tests could also be offered might be considered. The amount of conditions that this could be considered would be small. For the population as an entire, CF is that the obvious example and for ethnic groups, thalassemia is another example. Where screening programs of this sort exist and are well organized, the participation rate is usually high, indicating that they satisfy a real need.

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Received 22 July 2021; **Accepted** 26 July 2021; **Published** 31 July, 2021

How to cite this article: Sandhya Kille. "Population Genetic Screening Programmes." J Mol Biomark Diagn 12 (2021): 479.