Effectiveness, usefulness and ethical impact of screening tests in children in a cross-border setting

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STATUS
Accepted

PROPOSED BY

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SESSION DETAILS

Conference / Programme
ESOF2020 Trieste / ESOF 2020 Science Programme

Theme
Science and Society

Cross cutting approaches

Relevance of the selected approaches
The screening test are always a very debated clinical issue. Effective need, efficacy and clinical relevance are often a matter of discussion as well the costs of the procedures and the ethical issues. The building of a trans border guidelines for screening might be helpful to better understand the importance, the clinical impact and the relavance of screening tests

Target audience
Media **Policy makers** **General public** **Students**

Format / Duration
Interactive round table / 1h30

Abstract
Screening test is an exam that can identify an unrecognized disease in an apparently healthy and asymptomatic population. The most important screening tests are made on every newborn for certain metabolic, harmful or potentially fatal disorders that aren't otherwise apparent at birth. Although these

conditions are rare, early diagnosis and proper treatment sometimes can make the difference between lifelong impairment and healthy development. On the other hand, the primary immunodeficiency diseases, which are severe and potentially life-threatening conditions, do not have, at this time, national newborn screening tests. In some circumstances, the screening tests, although available, are not useful. This is the case of some genetic diseases with no treatment available and with high rate of mortality during childhood. For these conditions the carrier screening, used to determine if a person is a carrier for a specific genetic disease, seems to be more interesting. This kind of testing has been used by couples who are considering becoming pregnant to determine the risks of their child inheriting one of these genetic disorders. The carrier testing, accompanied by genetic counseling, has already been demonstrated to significantly reduce incidences of some genetic disorders like cystic fibrosis and spinal muscular atrophy. In this case, ethical issues are related to possible different severity among people with the same mutation, like in the case of spinal muscular atrophy. Whether the general population or just a specific population should be offered screening is also still controversial. Considering the different phenotype diseases, their prevalence, the availability of valid and cheap screening methods in the absence of national screening programs, we think that a new cross-border screening program could be planned to implement and to improve detection rate of neonatal curable diseases as well a carrier testing thus helping to compare strategies and ethical impact.

Special requirements

We intend to organize an interactive round table with 10 minutes time for each speaker and then a common general discussion will be applyed with interactive discussion and questions from audience.

Supporting documents

CV-short_engl.pdf (/uploads/1562494018-CV-short_engl.pdf)

CV of dr. Alberto Tommasini is enclosed

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