

Appropriate Means to Vulgarize the Human Cytogenetics

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Abstract

Purpose: Insufficient epidemiological data on the prevalence of chromosomal abnormalities may hamper investments in research and interventions for better prevention and treatment of these major threats to the population in numerous countries. The aim of this literature review is to contribute to the popularization of cytogenetics. **Methodology:** This literature review is a text which contains, analyzes and organizes several referenced articles in French and English which have been selected from electronic databases. It provides a documentation of various activities and progress that have an interest in the field of cytogenetics. **Results:** In the context of cytogenetics, it is necessary to establish partnerships to strengthen infrastructures and produce quality results. The setting up of cytogenetic services and incentives for participatory research give space to the community in the production of knowledge and innovation, alongside researchers. It's important to create biobanks, where samples of substances from the human body are collected for use in research into the causes and mechanisms of many diseases and their treatments. To carry out clinical trials aims at assessing the effectiveness of a therapeutic strategy, as is currently the case with the use of cytogenetic tests (karyotype, FISH, etc.) as tool of therapeutic decision and prognostic in cancer biology. The monitoring of the ethic of cytogenetic research projects allows protecting rights, security and welfare of subjects. To solve issues of data management

and analysis such as the obstacle to the acquisition of tools and the insufficiency in the training of researchers. Furthermore, dissemination of research results will lead to better understanding of research results, greater public engagement in science and greater social respect for research. **Conclusion:** Cytogenetics can benefit from these actions such as the development of its research infrastructures and training programs for its workforce as well as the development of its clinical care. All this would have an impact on the population's state of health.

Keywords

Human Cytogenetics, Vulgarization, Actions

1. Introduction

Cytogenetics is a discipline that studies genetic material organized in the form of chromosomes. It provides the basic elements on which strategies for clinical or biological diagnosis of chromosomal anomalies, screening, prevention, monitoring and treatment are developed, including genetic counseling for families [1] [2].

From its origins to the present day, cytogenetics has continued to progress in the analysis of its object of study, the human chromosome.

The first examination to analyze the chromosomes was the karyotype. New molecular cytogenetic technologies were developed which made it possible to identify chromosomal abnormalities down to a size of less than 5 megabases (MB) during the 1980s, then 1990s including fluorescence in situ hybridization (FISH) and the comparative genomic hybridization chip (array-CGH) [3].

It is well known that chromosomal disorders are a significant cause of premature death or often lifelong disability [4] [5] [6] [7].

Therefore, the results of literature review conducted in 2022 in Africa found that the proportion of chromosomal disorders among births with congenital anomalies was 8.94% ($p < 0.001$) [4]. Nevertheless, observations mention an underestimation of epidemiological data in underdeveloped countries mainly linked to the insufficiency of local registry systems and records of real cases [4] [5] [8] [9] [10]. This may hinder investments in research and interventions for prevention and better treatment of chromosomal abnormalities in many countries [4] [7].

In addition to this, health systems in developing countries are today faced with enormous challenges including the deficiency or absence of prenatal screening, cytogenetic diagnosis and associated services [4] [5] [7] [8]. There are also other factors such as the insufficiency and obsolescence of equipment, lack of qualified human resources and fundings [4] [8] [11]. All these problems therefore prevent the patient to benefit from adequate care and adapted genetic counseling [5] [7].

In view of the aforementioned difficulties, a question arises. How to improve the quality and quantity of human cytogenetics research?

The objective of this literature review is to contribute to the popularization of cytogenetics from a set of actions that are topical.

2. Methodology

The systematic literature review aims to provide an overview of various current activities and developments that are of interest in the field of human cytogenetics based on relevant literature. Starting from the objective, the investigations focused on several concepts:

- Roles of partnership in research,
- The place of cytogenetic services,
- Contributions of participatory research,
- Roles of biobanks,
- Interests of clinical trials,
- Advantages of monitoring the ethics of research projects,
- Adequate management and analysis of research data,
- And the importance of disseminating research results.

Thus, a documentary search of articles in French and English dating from 2021 to 2023 was undertaken to obtain an idea of all previous notions using electronic databases. The references of 37 selected publications were taken into account.

3. Results

The results of this work have been grouped in a table (**Table 1**) which displays the contributions of a certain number of current actions whose implementation of which helps to fill existing deficits or allows continued progression in the field of cytogenetics.

4. Discussion

This review identifies a number of current initiatives that would be of benefit to cytogenetics. These are analysed in this work to highlight the relevance of their implementation.

In low- and middle-income countries such as Africa, chromosomal abnormalities are likely to be underestimated due to the lack of accurate data on the causes of death [4]. Chromosomal disorders form a major category of genetic diseases [12]. They have a huge impact on overall health and well-being, causing multiple problems including mental retardation and/or physical disabilities [4].

However, most African economies allocate only scarce resources to the health research and private investment in this area is also modest. Thus, cooperation with developed countries will make help to strengthen research capacities by obtaining subsidies and producing quality results [13] [14].

The establishment of efficient and reliable cytogenetic services or laboratories constitutes the places for diagnosis of chromosomal abnormalities [5]. These

Table 1. Set of processes suitable for advancing cytogenetics [5] [12]-[37].

<u>Actions to be taken</u>	<u>Benefits</u>
<u>Development and maintenance of the partnerships</u>	<ul style="list-style-type: none"> ○ <u>Strengthens infrastructure</u> ○ Access to equipment specialized or to new sources of funding ○ Produces <u>quality results</u>
<u>Setting up cytogenetic services</u>	<ul style="list-style-type: none"> ○ Cytogenetic diagnosis: identification of chromosomal abnormalities of number and structural ○ Use of different techniques: karyotype, FISH <u>and</u> array-CGH ○ Gives the place to the community in the production of knowledge and Innovation alongside researchers
<u>To promote participatory research</u>	<ul style="list-style-type: none"> ○ <u>Members of communities can contribute to the design, the implementation and analysis of quantitative or qualitative research data</u> ○ Allows people to be heard on the issue of their own development
<u>Creation of biobanks</u>	<ul style="list-style-type: none"> ○ Collection of samples of substances from the human body (tissues, blood, DNA, cell pellets, etc.) associated with personal data on their donors ○ <u>Role</u> in researching the causes and mechanisms of disease as well as their treatments ○ Intended to serve scientific research ○ Increases sample availability
<u>Conduct of clinical trials</u>	<ul style="list-style-type: none"> ○ Aims to assess non-toxicity, tolerance and efficacy of a diagnostic method or a therapeutic strategy: cytogenetic tests for therapeutic decision and prognosis
<u>To monitor</u> the ethics of cytogenetic research projects	<ul style="list-style-type: none"> ○ Allows you to approve the conduct of research projects on subjects human beings and to periodically monitor its progress in order to ensure the protection of the rights of the subjects, as well as their safety and their well-being.
To solve <u>problems of data management and analysis</u> (example, obstacle to the acquisition of tools)	<ul style="list-style-type: none"> ○ <u>Provides the ability to describe, reduce, classify and clarify the data showing similarities or differences between variables or groups of variables</u>
<u>Dissemination of</u> research results (Books, Articles, Poster communications, Websites, etc.)	<ul style="list-style-type: none"> ○ Intended to attract the attention of governments or stakeholders on research results, giving a social, political or economic impact ○ Increase the visibility and understanding of research

structures offer tests, including karyotype, fluorescence in situ hybridization (FISH) and comparative genomic hybridization chip (array-CGH) in pre and postnatal. Chromosomal analysis techniques are performed from a variety of samples including amniotic fluid, chorionic villi, peripheral venous blood, bone marrow and lymph nodes [5] [15] [16].

In addition to previous actions, the development of participatory research is an approach that aims to conduct research by involving the community (associations, families, people with disabilities) and researchers from the study design to the analyze of data. Thus the level of community participation is attracting increasing interest in health science research [17] [18]. For example, associations actively are engaged in biological and clinical research. This commitment takes

various forms such as raising awareness of diseases, disseminating knowledge of pathologies and supporting doctoral students. Added to this co-financing of infrastructures and equipments, as well as participation in clinical trials [17] [19].

It is important to create biobanks that are places of systematic collections of human bodily samples (organs, tissues, blood and cells) as well as DNA as a carrier of genetic material [20].

The major objective of biobanks is to preserve these resources in optimal conditions in order to maintain their integrity and to make them available to the scientific community in general and in particular to facilitate the search for medicines and innovative diagnostic tests [21] [22] [23].

In the context of research, a clinical trial is a carefully designed prospective medical study that attempts to accurately answer a set of questions about the effects of a special treatment.

The WHO (World Health Organization) classification of haemopathies has included since 2001 into cytogenetic data [24]. Thus, in oncology, cytogenetic tests such as karyotype and FISH play a key role in diagnosis, prognosis, choice and monitoring of treatment [24] [25].

Research projects carried out on genetic diseases have made it possible to acquire new knowledge such as genetic testing, genetic counseling, prenatal diagnosis, assisted procreation technologies, embryo manipulation and the constitution of biobanks [23] [26]. They raise multiple ethical issues including the participation of minors in research, incidental findings, the protection of privacy and the interpretation of genetic findings in a personal or family referral. In addition, it is necessary to take into account the right to physical integrity but also the right to decide on the use of personal data [23] [26] [27]. In view of the above observations, it is necessary to emphasize that research protocols in the field of cytogenetics should be examined and approved by the local operational ethics committee.

Moreover, data represents information collected during the conduct of a research project. Their analysis and interpretation is a phase which essentially includes operations of classification, comparison, on each of the information collected in order to find its meaning and to draw conclusions [28] [29] [30]. In the area of research, there is a need for additional support in the management and analysis of large datasets as there seems to be a lack of knowledge about tools and resources for data management. Barriers to acquiring currently available tools are most often related to the financial burdens of small labs and unmet expectations for institutional support [28] [29] [30].

Institutions should facilitate the training of researchers in management strategies for conducting preserved data themselves, using for example secure networks and data encryption by user authentication through the password [28] [29] [30].

Dissemination of research is an integral part of the research process. It involves communicating research results to the target community, to decision-makers and others who may benefit from knowledge produced. Research released findings

leads to better understanding of findings, increased public engagement in science and greater social respect for research [31] [32].

There are several ways in which researchers communicate the results of their research. These include writing a book, a journal article or a communication aimed at the general public, and then organizing round tables. Other elements to consider include the creation of websites specific to the research project and the creation of mural or poster for public spaces. A researcher could also associate with others pursuing similar research objectives [32].

The interest of this study lies in the application of these measures which contribute to the benefits of cytogenetics in **Table 1**.

At the clinical level, the etiological diagnosis of chromosomal abnormalities makes thus it possible to specify the prognosis and set up appropriate medical follow-up then a systematic screening for complications. Appropriate genetic counseling on the risk of recurrence of the disease can also be established [33].

In the fields of teaching and research, the acquisition of laboratory materials and equipment allows to offer significant learning experiences to learners in universities and to develop research [34].

At national level, the above measures would enable the adoption of a prevention policy to limit the risks of spreading chromosomal abnormalities [35].

It is also desirable, as in some countries, to implement a legal and ethical framework, followed by legislation, for the practice of genetic counseling, pre or postnatal diagnosis. Should be added to previous list clinical trials, the creation of biobanks and prescriptions in the field of cytogenetics, with a view to protecting the rights of subjects, their safety and well-being [23] [26] [36] [37].

5. Conclusion

Cytogenetics is a clinico-biological discipline that is complemented and perpetuated by new research. Its funding could remain a priority in the government's economic recovery plan that is the development of its research infrastructures and training programs and for its workforce, as well as the development of its clinical care. This contributes to improving the life expectancy, quality of life and individual well-being.

Author's Contributions

ZPD collected the data. ZPD, AJLO, GFM, QDC, MOAB, PIA, BYEBG, BD, GVY, MIJMD have contributed to the data analysis.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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