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Contribution to the Struggle Against Chromosomal Abnormalities from Literature Review

Deh Zhou Patricia[1, *], Goulai Bi You Etienne Bazago[1], Montéomo Gnaté François[4], Coulibaly Quidana Désirée[1], Dieth Malika Joane Astrid[1], Okon Abou Joël Landry[1], Doukouré Brahima[2], Diomandé Mohenou Isidore Jean-Marie[1, 2], Yao Gnangoran Victor[3]

[1]Histology-Embryology-Cytogenetics Laboratory, Training and Research Unit-Medical Sciences, Félix Houphouët-Boigny University, Abidjan, Côte d'Ivoire

[2]Pathologycal Anatomy and Cytology Laboratory, Training and Research Unit-Medical Sciences, Félix Houphouët-Boigny University, Abidjan, Côte d'Ivoire

[3]Clinical Cytology and Reproductive Biology Laboratory, Training and Research Unit-Medical Sciences, Alassane Ouattara University, Bouaké, Côte d'Ivoire

[4]Physiology-Pharmacology-Pharmacopoeia Laboratory, Training and Research Unit-Nature Sciences, Nangui Abrogoua University, Abidjan, Côte d'Ivoire

Email address:

dehzpatricia@yahoo.fr (D. Z. Patricia)

[*]Corresponding author

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Abstract: Chromosomal abnormalities have a variable severity, which can range from lethality before birth to the need for permanent medical care. The objective of this work was to propose a strategy to the struggle against chromosomal abnormalities through Departments of cytogenetics from literature review. Several documents have been consulted on the activities of Departments (or Services) of cytogenetics in relation to health care, ethical, legal and social problems concerning chromosomal abnormalities. In addition there are teaching and international collaboration in the field of cytogenetics. The corresponding articles have been published in journals with editorial, reading and scientific committees. According to this literature review, Services of cytogenetics should emphasize appropriate technologies and affordable prices, which can reach a significant portion of the population. In these Services, genetic counseling respects the right to full information and possible solutions regarding chromosomal abnormalities. Prevention through community sensitization, screening of populations and the possibility of early diagnosis should be carried out. The teaching of cytogenetics is therefore the starting point from which programs to combat chromosomal abnormalities will be introduced. Departments of cytogenetics must be supported by regulatory structures constituted by the scientific, medical and non-professional communities. Research is an important component of medical cytogenetics as well as international collaboration. This work includes a set of coordinated actions whose implementation would deal with chromosomal abnormalities.

Keywords: Struggle, Chromosomal Abnormalities, Departments of Cytogenetics, Populations

1. Introduction

Cytogenetics is the branch of genetics that studies chromosomes and their abnormalities [1, 2]. The latter refer to number anomalies resulting from a supernumerary or missing chromosome and structural anomalies which involve one or more chromosomal breaks followed by abnormal reattachment [1]. Chromosomal abnormalities have a variable severity, which can range from lethality before birth to the need for permanent care [1, 3-7]. They

lead to dysmorphic syndrome, congenital malformations, intellectual retardation, infertility and then cancer [1, 3, 6, 7]. At least 7.5% of conceptions are affected by chromosomal abnormalities and most cause spontaneous abortions [8].

Departments (or Services) of cytogenetics provide care for patients with these chromosomal aberrations. These Services represent important sources of information on the frequencies of chromosomal abnormalities leading in-depth analyzes of these abnormalities [1, 3, 6, 7]. In addition these Services make it possible to determine the clinical profiles of patients referred for cytogenetic study and to highlight the variability of chromosomal abnormalities in different populations [3, 6, 7].

In Africa, most of the studies reviewed have shown that the available prevalences of chromosomal abnormalities are from hospital setting studies and more rarely on a population scale [3, 6, 7, 9, 10, 11]. Thus, it is difficult to collect accurate data on the prevalence of these medical conditions for the application of epidemiology to disease prevention and health promotion because many cases remain undiagnosed [3, 7].

In view of the seriousness of these medical conditions and their perceptible consequences in populations (reproductive failures, birth defects, medical dependence and significant emotional shocks to family and society), remains a question of major importance.

What are solutions or courses of action to fight against chromosomal abnormalities?

The objective of this work was to propose a strategy to the struggle against chromosomal abnormalities through Departments of cytogenetics from literature review.

2. Literature Review Methodology

Several documents have been consulted on the activities of Departments of cytogenetics in relation to health care, ethical, legal and social problems concerning chromosomal abnormalities. In addition there are teaching and international collaboration in the field of cytogenetics. The corresponding articles have been published in journals with editorial, reading and scientific committees.

3. Results

The various measures proposed in the fight against chromosomal abnormalities are presented in the diagram below.

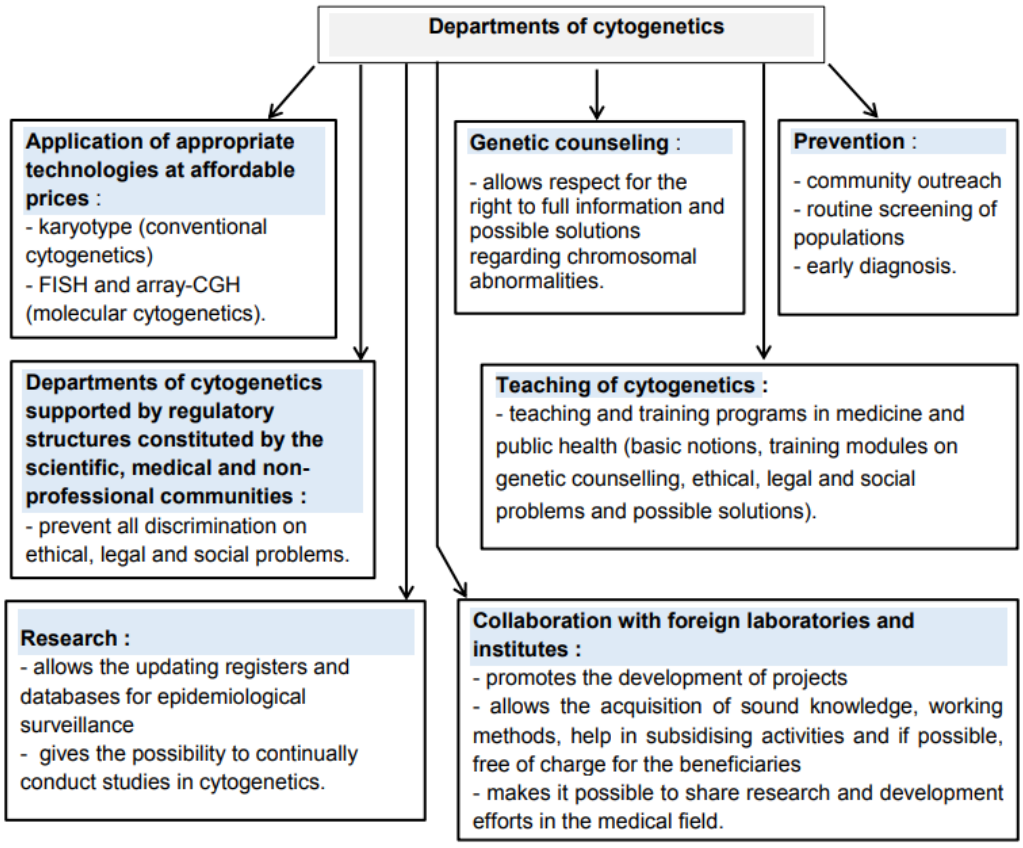

Figure 1. *Diagram summarizing the missions of Departments of cytogenetics in a strategy to struggle against chromosomal abnormalities.*

4. Discussion

The Departments of cytogenetics used to combat against chromosomal abnormalities should serve as a solid base from which to apply measures that make it possible to mitigate public health problems. Medical practitioners whose work there must obtain specialty degrees or equivalents such as the Certificates and Specialized Studies in cytogenetics, the Doctorate in cytogenetics and training courses [12].

This work carried out as part of the fight against chromosomal abnormalities, several missions have been assigned to the Departments of cytogenetics.

Services of cytogenetics should emphasize adequate technologies and affordable prices, which can reach a significant portion of the population.

Among the examples, the conventional cytogenetic technique represented by the human karyotype allows the observation and classification of chromosomes at the stage of metaphase and prometaphase of mitosis. In this method, banding techniques identify each chromosome pair by the pattern of light and dark bands [13-15]. The karyotype is indicated in newborns, children, adolescents and adults in constitutional pathology. The indications for karyotyping in acquired pathology are leukemia and malignant solid tumours [13]. The human karyotype being a morphological analysis, it remains a very time-consuming examination that cannot be automated. Moreover, when the chromosomal anomaly is very small, less than 5 megabases (Mb), it goes unnoticed on the karyotype [13, 16-19] and requires another type of diagnosis.

Molecular cytogenetics is of relatively recent appearance. Its main tools are two in situ hybridization techniques that allow the detection of chromosomal rearrangements of a size of less than 5 Mb [20].

Fluorescence in situ hybridization (FISH) makes it possible to locate the position and distribution of target sequences directly on the chromosomes using labeled probes consisting of the complementary sequences of these targets [17]. Fluorescent oligonucleotides are used as a probe.

Array-CGH or array comparative genomic hybridization is a fairly recent technique. It is based on the same principles as FISH, however, in the case of CGH, the probes are made from total genomic DNA [21].

While the standard karyotype detects a chromosomal abnormality in 9.5% of patients with mental retardation, numerous studies have shown that array-CGH can identify chromosomal (genomic) imbalances in 5 to 17% of patients, whose karyotype was previously considered normal [19, 22].

Genetic counseling is the process by which patients or relatives at risk of hereditary disease are advised and informed of the nature and consequences of this disease, the probability of developing or transmitting it to their offspring. It takes into account the options available to them in terms of life and family planning, so as to prevent illness or improve their situation [19, 23-25]. For example, chromosomes can be affected in their structure (deletion, duplication, translocation) [26-28]. These anomalies can be familial and raise the risk of recurrence within a family when one of the parents has no alteration in the phenotype and can transmit an unbalanced anomaly to his offspring [26].

Prevention in the field of health means acting in order to reduce the probability of the occurrence of a risk: accident, deficiency, disease, incapacity [29]. Prevention through community outreach, routine screening of populations and the possibility of early diagnosis is useful.

In fact, education of the population represents a guarantee for the success of any public health policy, especially since certain pathologies are directly linked to practices that are the basis of culture. In Africa, social or cultural risk factors such as consanguineous marriages [30] and a maternal age greater than 35 years [31], observed in certain communities, are likely to cause these anomalies.

Chromosomal rearrangements can occur at any age of a human being, predisposed or not. The consequences of these abnormalities strongly suggest routine screening of populations. For example, performing a karyotype can be justified in people with mental retardation, dysmorphic features, birth defects, sexual ambiguity and family history of chromosomal abnormalities. To these were added the people with repeated miscarriages and fertility problems. Before any assisted reproduction program, a karyotype may be requested to reduce the risk of failure, the cost being very high [3, 6, 7, 9, 32, 33].

To support this fight, regulatory structures formed by the scientific, medical and non-professional communities must preserve the dignity of the individual and his family. Then, these structures allow to prevent any discrimination concerning the ethical, legal and social problems associated with the fight against diseases, such as insurance or employment aspects.

It should be noted that prenatal diagnosis is an act of preventive medicine. The prenatal diagnosis represents all the techniques that make it possible to recognize a pathology more or less early, during intrauterine life in the embryo or fetus [34, 35]. The diagnosis made could interrupt or continue the pregnancy [34, 36]. It can be done either by morphological study using ultrasound or by biological examinations including karyotype, enzymatic assays and DNA study. Prenatal diagnosis should always be preceded by genetic counseling [37]. The hypothesis of the evocation of a termination of pregnancy, which is often refused by the parents, makes the use of prenatal diagnosis a difficult decision [38].

The teaching of cytogenetics [39] is therefore the essential starting point from which programs to combat genetic diseases and congenital malformations are introduced. In addition medical and public health education and training programs should include courses in cytogenetics and modules on genetic counselling. There are ethical, legal and social problems associated with these anomalies that need to be addressed.

Countries should also aim to provide continuing education opportunities for their health care professionals [40].

Research, which is an important component of medical cytogenetics, has been included in all the actions to be carried out. There is currently insufficient data available regarding the epidemiology of chromosomal abnormalities in developing countries. Updating effective surveillance systems (registers and databases) and continued investment in cytogenetic research are essential for the success of public health interventions, especially in regions where there are few resources [3, 7].

However, the poor performance of health systems in low-income countries in Africa also deserves special attention [41-43]. Collaboration with laboratories and institutes in

foreign countries that integrate various fields of cytogenetic research aims to promote the development of projects, the acquisition of solid knowledge, working methods as well as assistance in subsidizing activities and if possible, free for the beneficiaries. It is also about sharing research and development efforts in the medical field.

The interest of this work is to highlight through various arguments the missions attributed to Departments of cytogenetics in the context of the fight against chromosomal abnormalities. This reflection is essential, because it aims to help parents, family and society when affected people need to be diagnosed, informed and supported. Thus, the application of these measures gives the possibility to:

1. limit the risks of spreading chromosomal abnormalities,
2. adopt a national screening policy,
3. prospect of medically termination of pregnancy,
4. reduce mortality and especially morbidity related to these conditions,
5. involve psychologists, psychomotor therapists, physiotherapists, speech therapists and educators, then strengthen communication with parents so that they understand and adhere to this follow-up.

5. Conclusion

This study indicates the missions assigned to the Department of cytogenetics in the context of the struggle against chromosomal abnormalities. Cytogenetic diagnostic tools and genetic counselling as well as public awareness, could be integrated into care in general and in particular in areas where resources are scarce. The application of these measures will make it possible to limit the risks of propagation of these anomalies, reduce mortality and especially the morbidity linked to these diseases.

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