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Post Genomics: towards a Personalized Approach to Chromosome Abnormalities

Ivan Y Iourov*

Mental Health Research Center, Moscow, Russia; Russian National Research Medical University named after N.I. Pirogov, Separated Structural Unit "Clinical Research Institute of Pediatrics", Ministry of Health of Russian Federation, Moscow, Russia

Regardless of organizational and attitudinal difficulties, the application of post genomic technologies in personalized medicine provides a solid basis for individual molecular diagnosis and therapy with potential therapeutic benefits for genetic counseling. Generally, these opportunities are found considerable for complex and single-gene diseases [1,2]. However, probably due to the multilateral impact of chromosome abnormalities in the genomic context, post genomic technologies have been rarely used to address chromosomal syndromes and related cases. On the other hand, high-resolution genome-wide analysis of chromosome abnormalities demonstrates the possibilities of empirical and bioinformatic post genomic technologies to represent a basis for increasing the efficiency of the diagnosis and management [3-5]. Nevertheless, there is still no consensus on the way these developments can be used in genetic counseling.

Recently, a combination of molecular cytogenetic techniques with post genomic bioinformatic analyses has been shown as an effective way to genotype-phenotype correlations, elucidating disease mechanisms, and potential development of personalized molecular therapy in patients with chromosome abnormalities [4,5]. Moreover, similar approaches to chromosome aberrations based on *in silico* epigenomic, interactomic and meta bolomic analyses of molecular cytogenetic data allow developing a therapeutic strategy through proposing the pathogenic mechanism [5]. As a result, normalization of metabolic processes produced by chromosome abnormalities was proposed to be a way for treatments in at least a small proportion of cases of these presumably incurable genetic conditions.

Implementation of post genomics into clinical practice requires sophisticated roadmaps for organizational solutions despite convincible research data on appreciable success in personalized medicine obtained using post genomic technologies [6]. Furthermore, there are several types of genomic variations manifesting at chromosomal level (i.e chromosome abnormalities and instability, somatic mosaicism), which require additional research efforts to enhance corresponding post genomic approaches for the molecular diagnosis and correct clinical interpretation [7]. Still, state-of-the-art post genomic technologies for molecular cytogenetic analyses of chromosome abnormalities are able to shed light on disease mechanisms in chromosomal syndromes and cases of chromosome rearrangements for developing therapeutic strategies [3-7]. The potential of such emerging technologies increases, since new perspectives on treating at least a small proportion of chromosome abnormalities seem to exist. In conclusion, these achievements in post genomics clearly indicate that an important step forward towards a personalized approach to chromosome abnormalities has been made.

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***Corresponding author:** Ivan Y Iourov, Department of Medical Genetics, Russian Medical Academy of Postgraduate Education, Moscow 117152, Russia, Tel: 7-495-9528990; E-mail: ivan.iourov@gmail.com

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