The way to the next generation Reproductive genetics

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Reproductive genetics is a field that connects reproductive and genetic technologies, using all the achievements of genetics, with the aim of diagnosing and choosing the most appropriate treatment for infertility, as well as preventing the birth of a child with congenital anomalies and genetic diseases. The Human Genome Project was launched as a massive scientific effort to create a reference map of the human genome. Thanks to technological advances, genetic tests are becoming increasingly relevant in reproductive medicine, where they have three main purposes: identifying the causes of infertility; identification of genetic diseases transmitted to offspring; optimizing assisted reproductive technology (ART). The standard genetic testing algorithm for infertility research currently includes chromosomal analysis, microdeletions of the Y-chromosome, and molecular genetic studies of certain mutations, such as congenital thrombophilias and CFTR. Strategies based on NGS (next-generation sequencing) offer the opportunity not only to optimize genetic testing in reproductive medicine (since in one step it may be possible to analyze potential causes of infertility, carry out carrier screening and to support ART), but also to adapt the therapeutic solution to the specific genomic characteristics of the patients. The challenge for future medicine is to move from a population-based approach to an individual-based approach. New technologies are driving this revolution. In recent years, new single-cell genomics methods have emerged that have revolutionized our ability to identify and characterize the cells that make up complex tissues. The Human Cell Atlas (HCA) project was launched as an international collaborative effort to create comprehensive reference maps of all human cells – the basic units of life – as a basis for both understanding human health and diagnosing, monitoring and treatment of diseases. Using advances in sequencing technologies, this project has the ambitious goal of profiling cellular transcriptomes, along with their spatial determination, to map the identified cell types onto the tissue architecture of the human body. This information will give an unprecedented impetus to the development of all medicine.