
Preface

In April 2011, the website *The DNA Exchange* ran a story about the origin of our convention of referring to the short and long arms of chromosomes as “p” and “q.” Several possible explanations for how this usage came into being were presented in a somewhat whimsical manner.

Did we really go with p from the French *petite* and q because it alphabetically follows p? Was there really a “French vs. English” argument? Was it supposed to be p and g (from the French *grande*) but changed due to a typesetting error? Was Hardy-Weinberg equilibrium ($p+q=1$) invoked?

This prompted a flurry of comments over the Listserv used by cytogeneticists. Ultimately, several participants of the 1966 “Chicago Conference” weighed in, and Dr. Kurt Hirschhorn, who chaired the session at that conference, confirmed that the decision to go with p and q resulted from a combination of (sometimes spirited) debate, compromise (p really is for *petite*), logic, and, yes, agreement that $p+q=1$.

This is all great fun. But the story in *The DNA Exchange* also spawned other comments. It opened with:

Karyotypes are sooooo 20th century. Time was when a ripe crop of G-banded chromosomes promised a fruitful harvest of genetic secrets. But nowadays a Giemsa-stained karyotype seems like a quaint low resolution black and white TV set – those cute little D & G groups even have rabbit-ear antennas – compared with the bright, sexy colors of FISH, the fine oligonucleotide detail of microarrays, and the dense volumes of data of generated by high throughput DNA sequencing.

Some cytogeneticists took offense at this.

People have been predicting the demise of cytogenetics for decades; this tended to happen each time new technology, such as DNA analysis or fluorescence *in situ* hybridization, became available. And yet we are still here.

Interestingly, this idea was significant as the previous edition of this book went to press in 2005 due to the increasingly important role of many FISH assays. In the preface to that edition, we discussed that while some classically trained cytogeneticists were concerned that FISH was going to put them out of work, Dorothy Warburton had predicted, years earlier, that FISH would actually provide the cytogenetics lab with an even more important diagnostic and prognostic role. She was of course correct.

Now we have microarrays. This edition of our book has a chapter dedicated to this technology, and several authors also deal with it in their individual chapters. The term “cytogenomics” (chromosome analysis using molecular techniques) is working its way into our lexicon.

Once again, there is talk, if not concern, that arrays could mean the unemployment line for cytogeneticists and, if not arrays, then perhaps next-generation sequencing. And once again, Dorothy put things into perspective:

The way I look at it is that cytogenetics is not about a technique, but a field of knowledge. We may change the way we look at chromosomes, but the questions and problems remain the same. A technique is only as good as our ability to interpret what we see in a way that helps families, and having molecular training does not provide the experience necessary to do this. We would never have known about bal-

anced translocations without looking at chromosomes, but now we have a way to tell if they are really balanced or not. I also believe that we will never be able to stop using chromosome preparations to interpret what we see on arrays. We have many examples where confirming array data has revealed unexpected kinds of rearrangements, as well as mosaicism. These are things that have much more significance for counseling than a simple call of a dup or del. I don't believe sequencing will change this.

I was first advised to find another field in 1969 (right before banding). So far I still have a job, although what I look at day to day has changed a great deal. "Classical" is pretty much a synonym for "in the past," so yes, classical cytogenetics may no longer be practiced. However, what is here is exciting and challenging and requires every technique in our playbook.

This third edition of *The Principles of Clinical Cytogenetics* was prompted by significant advances in the field since the last edition of this book was published. So while it is true that the way we look at chromosomes will likely continue to evolve, we do not expect to stop looking at them any time soon.

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