“Gardner and Sutherland’s chromosome abnormalities and genetic counseling” by R.J. McKinlay Gardner, David J. Amor. Oxford University Press

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As a classically trained cytogeneticist who has spent the majority of my career to date reporting microarray results (and having spent the last few months learning to become proficient at sequence variant interpretation), I welcomed the opportunity to return to cytogenetics and review a book that is such an integral part of the laboratory.

The changes to this, the fifth, edition are noticeable even from the contents page, with sections now grouped to reflect the clinical contexts around which many laboratories and services are now organized, reflecting the change from distinguishing between cytogenetic and molecular genetic testing to genomic and genetic testing depending on the referral reason.

Perhaps unsurprisingly, my favorite change to this book is the normal chromosomal variants chapter. It is reassuring to see the recurrent CNV’s and the papers we use to inform interpretation become so well-established that they are now in a ‘text book’ rather than research and literature. There is also a really good description of the resources available for interpretation including DECIPHER, DGV and ClinGen. The removal of the Fragile X chapter again reflects current practice and that FRAX is now commonly a second-line test.

The reproductive failure and prenatal diagnosis chapters of previous editions have joined to form the ‘reproductive

cytogenetics’ section. The most notable amendment to this section being the change from ‘Prenatal Diagnostic Procedures’ to ‘Prenatal Testing Procedures’. This subtle but critical change has been necessitated by the incorporation of NIPT into routine clinical practice as a screening test and therefore its rightful inclusion in this chapter. Previously, NIPT had been restricted to several paragraphs under the banner of ‘Experimental Approaches’. This, more than most other examples, portrays how quickly genetics and genomics has evolved and changed over a relatively short period of time. The information on NIPT flows nicely into the ‘screening for fetal trisomy’ information followed by ‘fetal ultrasonographic anomalies’. Accordingly, the section on ‘microarray-level rearrangements’ has also been updated to include recent references to large studies reflecting on the impact of the use of microarrays in prenatal testing. The description of mosaicism now includes the contribution of techniques such as QF-PCR and microarrays also.

In summary, this new version has maintained Gardner and Sutherland as ‘the’ essential text for laboratory genetics. The changes have made the book current, relevant, reflective of current practice and hugely user-friendly and “hands on”.

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