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BOOK REVIEWS

So that is why you stink!

Meaningful scents around the world. Olfactory, Chemical, Biological, and Cultural considerations

Roman Kaiser
J Wiley VCH 2006.
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The plant world enhances our lives with wonderful fragrances, or sometimes stinks up our environment with a vast array of volatile organic molecules. Humans, whose sense of smell has evolved over time, are still able to perceive at least 1000 different molecules activate some 1100 odorant receptors, the single largest gene family in our genome.

As someone who has a specific odour blindness, in this case, to my wife's favorite daytime lily, *Acididanthera* and, as a practicing clinical geneticist, I volunteered to review this book, in part as a personal journey of discovery, but also to bring it to the attention of graduate students, post-docs, and young clinical investigators in search of the next frontier in clinical research (Figure 1).

The author, Roman Kaiser, is a chemist with a 30-year involvement in natural scent research on behalf of Givaudan Schweiz AG, the Swiss-based company with a 30% share of the world's perfume market.

This book is many things and will appeal to a diverse audience. For the most part, it is both a personal travelog and encyclopedia, which brings together the olfactory, chemical, biological, and cultural considerations of scents.

Kaiser has perfected a simple method of collecting a high yield of scents in the field. He describes the design and technical details of an absorption trap, which makes use of a battery-operated pump and a variety of hollow-stemmed goblet-like glasswares with which he can extract up to 200μg of pure scent for subsequent analysis using combined gas chromatography/mass spectroscopy.

Figure 1 Acidanthera murielae

The author has enjoyed a long collaboration with Pro-Natura International, a well-known non-profit rain forest preservation group who provided access to their floating laboratory – a dirigible with a suspended platform that provides a means of direct access to various plants in the forest canopy of the lower Amazon, Gabon and Madagascar. His other expeditions took him to the biotopes of the Ligurian Coast, Italy; Papua, New Guinea, Indonesia; and the highlands of Yunnan province in China. The reader is treated to a travelog illustrated with an almost extravagant use of color photographs and maps along with an occasional recipe. Details of the chemical structure of 232 individual chemical molecules are included along with a 52-page technical chapter listing in detail the composition of the 110 scents discussed in the book.

The problem as to how the brain represents the external world is at the very center of art and the very core of philosophy, psychology, and neuroscience. These sentiments are drawn from the Nobel Lectures of Richard Axle and Linda Buck who won the prize in Physiology and Medicine in 2004. In the following year, the National Institute of Deafness and Other Communication Disorders, one of the National Institutes of Health, spent approximately 45 million dollars on studies involving the sense of smell.

To me, these events signal an exciting opportunity for clinical research into the largely unexplored field of the effect of mutation in odor receptor genes and human behavior.

Two large supergene families of G protein-coupled receptors have been identified as pheromone receptors in the vomeronasal organ of the mammalian brain. While it is true that in humans, two-thirds or more of these genes are dysfunctional owing to gene degradation, those remaining are thought to play a role in the mammalian social systems, including sex, reproductive status, maternal newborn recognition, competitive ability, and health status.

This book offers a wonderful overview of the biochemistry of scents and is a good

place to start if you are looking for an introduction to the next frontier in neuroscience. Many of these odorant receptor genes are located in the telomeric region of almost all chromosomes and are seen in abundance on chromosomes 7, 16, and 17. A number of our patients are now being identified with telomeric deletions. Is there a connection between odor receptor deletions and the developmental phenotype of these patients?

If you are going to be around Nice anytime soon, you might want to plan a day trip to Le Manie, 80 km east along the coast. It has been described as being one of the most scented regions of our world.

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Molecular bases of mental retardation

Mental Retardation and Developmental Delay – Genetic and Epigenetic Factors

Moyra Smith (ed)
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It was a great pleasure reviewing this book owing to its practical and comprehensive way of presenting such a complex material, reflecting 40 years' experience in the field of genetics, biochemistry and inborn errors of metabolism. The book is addressed to those interested in molecular mechanisms underlying embryonic nervous system development, as well as mechanisms that lead to structural and functional brain abnormalities.

The material is structured in 10 chapters covering different aspects of mental retardation and developmental delay. All chapters are very dense, starting with a few clinical data, followed by detailed, yet comprehensive information regarding molecular mechanisms for the most important specific disorders. The text is appropriately illustrated with schematic diagrams, which make it very easy to follow.

The introductory part discusses the key events in understanding the causes of mental retardation – all the social, environmental, metabolic and genetic factors are presented.

Neurogenesis and neuronal migration, maturation and function are discussed from the molecular point of view in the second chapter. Although the subject seems difficult, the author has achieved an outstanding presentation of the mechanisms underlying normal and abnormal development for each step separately.

The subsequent four chapters deal with the main categories of defects associated with mental retardation: structural brain anomalies, dysmorphic features (specific syndromes), other neurological defects (eg seizures) and metabolic defects. Each of them is a state of the art presentation in the field. I was very pleased to discover that some of the most recently described disorders (eg Mowat–Wilson's syndrome) have been included in the presentation, as well as the newest molecular data for every category of mental retardation, reflecting the vast experience of the author. The text combines molecular biology, genetics, metabolic pathways and clinical practice, but the focus on molecular data is more than obvious. The rich selection of illustrations (metabolic reactions and tables) makes the material practical and instructive. Once you start reading this book, you cannot put it away – you become more and more eager to discover something new about the next disorder.

Autism is another part to be mentioned, especially because this is such a difficult field to manage in practice. The complex data regarding autism inheritance are clearly and simply presented, showing once more the experience of the author.

The chapter referring to functional genomics and epigenetics is remarkable. Many of the notions explained here have been recently introduced and not fully understood, but the author presents them in a very clear and comprehensive way.

Practitioners should not miss the last two chapters, discussing the necessary information for a correct diagnosis, prevention and treatment directions. They will appreciate the global assessment recommendations to establish the diagnosis, as well as the multitude of therapeutic approaches presented here.

Another particularly useful aspect for quick reference is the 'Index' – detailed and accurate – very good, when you look for a specific gene or metabolic pathway.

The bibliography is remarkable – 53 pages containing almost 1000 articles, most of them recent and very interesting.

In conclusion, this is an excellent book that should be on all pediatric and neurologic wards, medical genetics centers as well as medical school libraries. The material is dense, but provides you with the most updated data in the field■

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