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Gnrique Mdicale Enseignement Thmatique Collectif

Joacim Rocklöv



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Génétique médicale CNEPGM (Collège National des Enseignants et Praticiens de Génétique, Caroline Schluth-Bolard, Martin Krahn, Damien Sanlaville, 2022-08-30) Conforme au programme du Diplôme de formation générale en sciences médicales DFGSM 2-3 cet ouvrage apporte les connaissances fondamentales en génétique médicale. Tous les items relatifs à la discipline sont abordés en 32 chapitres. Après une introduction détaillée sur l'architecture, la structure et la fonction du génome humain, l'ouvrage s'articule autour de quatre grands axes : génétique formelle, génétique chromosomique, génétique moléculaire, génétique et pratique médicale. La dernière partie est consacrée aux maladies rares et maladies génétiques qui sont au programme des ECN-EDN. Cette 2^e édition procède à l'ajout et la mise à jour de l'ensemble des chapitres et introduit de nouveaux chapitres sur la génétique des maladies complexes, la médecine génomique et les maladies rares. Le propos est clair et didactique, appuyé de plus de 160 schémas, illustrations et tableaux tout en couleurs. Le cours est complété par de nouveaux QCM, QCS, QRU et QROC corrigés en fin d'ouvrage permettant de tester ses connaissances. Des compléments numériques permettent de télécharger l'intégralité des illustrations et donnent accès à deux textes sur le conseil génétique. Ce livre s'adresse tout particulièrement aux étudiants de DFGSM 2-3, Médecine aux étudiants en médecine ou en pharmacie et aux étudiants en Licence et Master de biologie. Il constitue également un livre de référence pour tout étudiant en DFGSM pour la préparation des items de génétique au programme des ECN-EDN ainsi que pour tout interne débutant la spécialité de génétique médicale. Il intéressera enfin les sages-femmes, infirmières et conseillers en génétique ainsi que tout praticien ou interne dont la spécialité présente un attrait pour la génétique médicale. Laboratoire sous la gérance du Collège national des enseignants et praticiens de génétique médicale. Cet ouvrage a été coordonné par Martin Krahn, Damien Sanlaville et Caroline Schluth-Bolard. Il est le fruit du travail collectif de près de 80 enseignants et praticiens exerçant tous leurs activités médicales d'enseignement ou de recherches dans le domaine de la génétique clinique, chromosomique ou moléculaire. Génétique médicale, 2022 **Bulletin critique du livre français**, 2005

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