

release from the United States Centers for Disease Control, summarizing statistical data related to birth and death rates, congenital anomalies, causes of death, etc.]

Cystic Fibrosis and Inborn Errors of Metabolism

- Cutting GR: Modifier genes in Mendelian disorders: the example of cystic fibrosis. *Ann N Y Acad Sci* 1214:57-69, 2010. [An outstanding review on modifier genes in so-called monogenic disorders, using cystic fibrosis as a template.]
- Farrell PM, Rosenstein BJ, White TB, et al: Guidelines for diagnosis of cystic fibrosis in newborns through older adults: Cystic Fibrosis Foundation consensus report. *J Pediatr* 153:S4-S14, 2008. [A somewhat dated, but gold standard consensus report on diagnostic criteria for cystic fibrosis, including variant forms.]
- Mitchell JJ, Trakadis YJ, Scriver CR: Phenylalanine hydroxylase deficiency. *Genet Med* 13:697-707, 2011. [A straightforward review on this prototype Mendelian disorder; useful if reader is seeking additional information beyond current chapter.]
- Ramsey BW, Davies J, McElvaney NG, et al: A CFTR potentiator in patients with cystic fibrosis and the G551D mutation. *N Engl J Med* 365:1663-72, 2011. [An original research article describing one of the first therapeutic strategies in cystic fibrosis that works through potentiating the function of CFTR protein.]
- Ratjen F, McColley SA: Update in cystic fibrosis 2011. *Am J Respir Crit Care Med* 185:933-6, 2012. [A clinically oriented review on cystic fibrosis that discusses many of the longer term sequela contributing to morbidity and mortality.]

Diseases of Prematurity and SIDS

- Casteels I, Cassiman C, Van Calster J, et al: Educational paper: Retinopathy of prematurity. *Eur J Pediatr* 171:887-93, 2012. [A well rounded review on retinopathy of prematurity, which includes a discussion of how our understanding of the pathophysiology of this entity has evolved over time.]
- Gien J, Kinsella JP: Pathogenesis and treatment of bronchopulmonary dysplasia. *Curr Opin Pediatr* 23:305-13, 2011. [An outstanding review

on bronchopulmonary dysplasia, which like the preceding reference, discusses how enhancements in understanding the pathophysiology of BPD have impacted prevention and treatment strategies.]

- Gower WA, Noguee LM: Surfactant dysfunction. *Paediatr Respir Rev* 12:223-9, 2011. [A review discussing the genetic defects associated with surfactant dysfunction, and associated acute or chronic pulmonary disorders.]
- Kinney HC, Thach BT: The sudden infant death syndrome. *N Engl J Med* 361:795-805, 2009. [A highly relevant review on SIDS, from one of the pioneering researchers elucidating the neuropathology of this condition.]
- Neu J, Walker WA: Necrotizing enterocolitis. *N Engl J Med* 364:255-64, 2011. [An outstanding review on this condition from one of the leading physician scientists in this field; particularly exemplary photomicrographs and illustrations.]

Tumors of Infancy and Childhood

- Chau YY, Hastie ND: The role of Wt1 in regulating mesenchyme in cancer, development, and tissue homeostasis. *Trends Genet* 28:515-24, 2012. [A comprehensive treatise on role of the Wt1 protein in development and in cancer.]
- Hamilton TE, Shamberger RC: Wilms tumor: recent advances in clinical care and biology. *Semin Pediatr Surg* 21:15-20, 2012. [A review on Wilms tumor that discusses underlying genetic susceptibility, natural history, and treatment options.]
- Maris JM: Recent advances in neuroblastoma. *N Engl J Med* 362:2202-11, 2010. [A well rounded review on neuroblastomas.]
- Molenaar JJ, Koster J, Zwijnenburg DA, et al: Sequencing of neuroblastoma identifies chromothripsis and defects in neurogenesis genes. *Nature* 483:589-93, 2012. [An original research article describing two new classes of recurrent genetic alterations in neuroblastomas.]
- Weksberg R, Shuman C, Beckwith JB: Beckwith-Wiedemann syndrome. *Eur J Hum Genet* 18:8-14, 2010. [A review by Dr. Beckwith on the eponymously named syndrome that predisposes to several pediatric neoplasms, including Wilms tumor.]