

REFERENCES

1. **Aase JM(1990).** *Diagnostic Dysmorphology*. 3rd ed. New York, NY: Plenum Medical Book Co; 32 –34.
2. **Accardo PJ , Capute AJ (1994).** Mental Retardation. In: F Oski, CD DeAngelis and RD Feigin *et al.*, Editors, *Principles and Practice of Pediatrics*, Lippincott, Philadelphia pp. 673–679.
3. **Albertson DG, Pinkel D(2003).** Genomic micro arrays in human genetic disease and cancer. *Hum Mol Genet.* 12 (spec No. 2):R145–R152.
4. **Alexander K.C. Leung, M.B.B.S (1999).** Alberta Children's Hospital and University of Calgary, Alberta, Canada.
5. **Al-Qudah AA (1998).** Screening for congenital hypothyroidism in cognitively **delayed** children. *Ann Trop Pediatr* . 18: 285–288.
6. **American Academy of Pediatrics, Committee on Committee on Children With Disabilities(2006).** Developmental surveillance and screening of infants and young children. *Pediatrics* 108 :192.
7. **American Academy of Pediatrics Committee on Children with Disabilities(2001).** Developmental surveillance and screening in infants and young children. *Pediatrics* . 108: 192–196.
8. **American Psychiatric Association (2000).** Diagnostic and statistical manual of mental disorders, text revision, 4th ed. Washington: American Psychiatric Association ; 41–49.
9. **Amir N .; El-Peleg O.; Shalev, R. S.; et al(1987) :** Glutaric aciduria type I: clinical heterogeneity and neuroradiologic features. *Neurology* 37: 1654-1657.
10. **Ankaran S, Laptook AR, Ehrenkranz RA, et al (2005).** "Whole-body hypothermia for neonates with hypoxic-ischemic encephalopathy". *N. Engl. J. Med.* 353 (15): 1574–84.

11. **Ashwal S, Russman BS, Blasco PA, et al (2004).** "Practice parameter: diagnostic assessment of the child with cerebral palsy: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society". *Neurology* **62** (6): 851–63.
12. **Asku F(1990).** Nature and prognosis of seizures in patients with cerebral palsy. *Dev Med Child Neurol* 32:661–668.
13. **Bachman JA, Bachman WG, Franzel AS, et al (1994)**
Preteaching developmentally delayed preschoolers to aid vision screening. *Optom Vis Sci* . 71: 713–716.
14. **Bacino CA, Kashork CD, Davino NA, et al (2000)** Detection of a cryptic translocation in a family with mental retardation using FISH and telomere region-specific probes. *Am J Med Genet.* ;92 :250 –255.
15. **Balaban B, Yasar E, Dal U, et al (2007).** "The effect of hinged ankle-foot orthosis on gait and energy expenditure in spastic hemiplegic cerebral palsy". *Disability and rehabilitation* **29** (2): 139–44.
16. **Banatvala JE, Brown DW(2004);** Rubella.; *Lancet* 3;363(9415):1127-37.
17. **Battaglia A, Bianchini E, Carey JC (1999).** Diagnostic yield of the comprehensive assessment of developmental delay/mental retardation in an institute of child neuropsychiatry. *Am J Med Genet* . 82: 60–66.
18. **Bennett FC, Guralnick MJ(1991).** Effectiveness of developmental intervention in the first five years of life. *Pediatr Clin North Am* 38:1513-1528.
19. **Berrueta-Clement JR, Schweinhard L, Barnett W, et al (1984).**
Changed lives: the effects of the Perry preschool program on

- youths through age 19. No. 8 of Monographs of the High/Scope Educational Research Foundation. Ypsilanti, Mich.: High/Scope Press.
20. **Beukelman, David R.; Mirenda (1999).** Augmentative and Alternative Communication: Management of severe communication disorders in children and adults. Pat (2 ed.). Baltimore: Paul H Brookes Publishing Co. pp. 246–249.
 21. **Biesecker LG(2002).** The end of the beginning of chromosome ends. *Am J Med Genet.* 107 :263 –266.
 22. **Blackman JA (1991).** Neonatal intensive care: is it worth it? Developmental sequelae of very low birthweight. *Pediatr Clin North Am* 38:1497-1511 .
 23. **Blasco PA.(1991)** Pitfalls in developmental diagnosis. *Pediatr Clin North Am* 38:1425-1438.
 24. **Bouhadiba Z, Dacher J, Monroc M, et al(2000).** MRI of the brain in the evaluation of children with **developmental delay**. *J Radiol .* 81: 870–873.
 25. **Brandt, N. J.; Brandt, S.; Christensen, et al(1978) :** Glutaric aciduria in progressive choreo-athetosis. *Clin. Genet.* 13: 77-80.
 26. **Cakmak A, Zeyrek D, Cekin A, Karazeybek H (2008).** Dandy-Walker syndrome together with occipital encephalocele. *Minerva Pediatr.* 60(4):465-8.
 27. **Camfield CS, Camfield PR, Wirrell E, et al(1996).** Incidence of epilepsy in childhood and adolescents: a population based study in Nova Scotia from 1977–1985. *Epilepsia* 37:19–23.
 - 28 **Capute and Accardo's(2005).** Neurodevelopmental.28 Disabilities in Infancy and Childhood, *Third* edition.

29. **Capute AJ, Accardo PJ(1996).** A neurodevelopmental perspective on the continuum of developmental disabilities. In: Capute AJ, Accardo PJ, eds. *Developmental Disabilities in Infancy and Childhood* 2nd ed. Vol 1. Baltimore, MD: Paul H. Brookes; 1–22
30. **Centeno RS, Winter J, Bentson JR, et al (1983):** CT evaluation of *Haemophilus influenzae* meningitis with clinical and pathologic correlation. *Comput Radiol* 7(4):243-249.
31. **Chelly, J., & Mandel, J (2001).** L. Monogenic causes of X-linked mental retardation. *Nature Reviews Genetics* 2, 669–679 .
32. **Chow SL, Rohan C, Morris AA. (2003).** "Rhabdomyolysis in glutaric aciduria type I." *J Inherit Metab Dis.* 26 (7): 711–2.
33. **Coplan J. ELM Scale(1983)** (Early Language Milestone Scale). Tulsa, Okla.: Modern Educational Corporation.
34. **Couvert P, Bienvenu T, Aquaviva C, et al (2001).** MECP2 is highly mutated in X-linked mental retardation. *Hum Mol Genet.* 10 :941 –946
35. **Craigien, W. J.(1996) :** Leigh disease with deficiency of lipoamide dehydrogenase: treatment failure with dichloroacetate. *Pediat. Neurol.* 14: 69-71.
36. **Crawford DC, Acuna JM, Sherman SL(2001).** FMR1 and the fragile X syndrome: human genome epidemiology review. *Genet Med* .3: 359–371.
37. **Curatolo P, Arpino C, Stazi MA, et al (1995).** Risk factors for the co-occurrence of partial epilepsy, cerebral palsy and mental retardation. *Dev Med Child Neurol* 37:776–782.
38. **Curry CJ, Stevenson RE, Aughton D, et al (1997).** Evaluation of mental retardation: recommendations of a consensus conference—

- American College of Medical Genetics. Am J Med Genet. 72 :468–477.
39. **Dart, Hurlbut, Boyer-Hassen (2004)** pp. 1Timbrell, J.A., ed . "Biochemical mechanisms of toxicity: Specific examples". Principles of Biochemical Toxicology, 4th edition. Informa Health Care.
40. **Demaerel P, Kingsley DP, Kendall BE(1993)**. Isolated neurodevelopmental delay in childhood: clinicoradiological correlation in 170 patients. *Pediatr Radiol* . 23: 29–33.
41. **Di Mario S, Basevi V, Gagliotti C, et al(2009)**; Prenatal education for congenital toxoplasmosis. Cochrane Database Syst Rev 21;(1):CD006171.
42. **Dionisi-Vici C, Deodato F, Raschinger W, et al (2006)**. "Classical organic acidurias, propionic aciduria, methylmalonic aciduria, and isovaleric aciduria: long-term outcome and effects of expanded newborn screening using tandem mass spectrometry". J Inherit Metab Dis. 29 (2-3): 383–389.
43. **Dobos AEJ, Dworkin PH, Bernstein BA (1994)**. Pediatricians' approaches to developmental problems: has the gap been narrowed? Dev Behav Pediatr . 15: 34–38
44. **Dworkin PH (1989)**. British and American recommendations for developmental monitoring: the role of surveillance. Pediatrics 84:1000-1010.
45. **Fenichel GM (2001)**. Psychomotor retardation and regression. In: Clinical pediatric neurology: a signs and symptoms approach, 4th ed. Philadelphia: WB Saunders, 117–147.
46. **Filiano JJ, Bellimer SG, Kunz PL(2002)**. Tandem mass spectrometry and newborn screening: pilot data and review. Pediatr Neurol. 26 :201 –204

- 47.**First LR, Palfrey JS(1994).** The infant or young child with developmental delay. *N Engl J Med* . 330: 478–483
- 48.**Folsom RC, Diefendorf AO (1999).** Physiologic and behavioral approaches to pediatric hearing assessment. *Pediatr Clin North Am* . 46: 107–120.
- 49.**Franceschetti S, Antozzi C, Binelli S, et al(1993).** Progressive myoclonus epilepsies: an electroclinical, biochemical, morphological and molecular genetic study of 17 cases. *Acta Neurol Scand* . 87: 219–223.
- 50.**Frankenburg WC, Dodds J, Archer P, et al (1992)** The Denver II: a major revision and restandardization of the Denver Developmental Screening Test. *Pediatrics*. 89 :91 –97.
- 51.**Frankenburg, W.K. and Dodds, J.B(1967).** The Denver Developmental Screening Test. *J. Pediat.*, 71:181.
- 52.**Frankenburg, W.K., Dodds, J., Archer, P. et al(1992).** The DENVER II: A major revision and restandardization of the Denver Developmental Screening Test. *Pediatrics*, 89:91-97, 1992
- 53.**Frankenburg, William K. (2002).** "Developmental Surveillance and Screening of Infants and Young Children". *Pediatrics* **109** (109): 144–145.
- 54.**Gabrielli O, Salvolini U, Coppa GV, et al (1990).** Magnetic resonance imaging in the malformative syndromes with mental retardation. *Pediatr Radiol*. 21 :16 –19
- 55.**Ghalli I et al.(2002).** Egyptian growth curves for infants, children and adolescents. In: Proceedings of the 1st National Congress for Egyptian Growth Curves, Cairo University, 11 December 2003. Cairo, Department of Bioanthropology, National Research Centre.

56. **Gilbert GL;(2002).** Infections in pregnant women.; Med J 4;176(5):229-36.
57. **Ginsberg L (2004).** "Difficult and recurrent meningitis". Journal of Neurology, Neurosurgery, and Psychiatry 75 Suppl 1: i16–21.
58. **Gkoltsiou Konstantina , Meropi Tzoufi, Serena Counsell, et al (2008) .** Serial brain MRI and ultrasound findings: Relation to gestational age, bilirubin level, neonatal neurologic status and neurodevelopmental outcome in infants at risk of kernicterus. *Early Human Development* . 11 829-838.
59. **Glascoc, Frances Page et al. (1992).** "Accuracy of the Denver-II in Developmental Screening". *Pediatrics* (89): 1221–1225.
60. **Gonzalez, Jason; Willis, Monte S(2010).** title=Ivar Asbjorn Folling Discovered Phenylketonuria (PKU) - journal=lab medicine - volume=41 - number=2 - pages=118-119
61. **Goodman, S. I.; Markey, S. P.; Moe, P. G et al (1975).** Glutaric aciduria; a 'new' disorder of amino acid metabolism. *Biochem. Med.* 12: 12-21, 1975.
62. **Guthrie R(1986).** Screening for "inborn errors of metabolism" in the newborn infant—a multiple test program. *Birth Defects* 4:92-98.
63. **Guthrie R, Susi A(1963).** A simple phenylalanine method for detecting phenylketonuria in large populations of newborn infants. *Pediatrics* 32:338-343.
64. **Harber JS (1991).** Early diagnosis and referral of children with developmental disabilities, *Am Fam Pract* **43** pp. 132–140.

65. **Hauser WA(1994).** The prevalence and incidence of convulsive disorders in childhood. *Epilepsia* 35 [Suppl 2]:S1–6.
66. **Hauser WA, Hesdorffer DC (1990).** *Epilepsy, Frequency, Causes, and Consequences.* New York : Demos,18–21.
67. **Henderson HE, Goodman R, Schram J, Diamond E, et al (1981)** Biochemical screening for inherited metabolic disorders in the mentally retarded. *S Afr Med J* . 1981; 60: 731–733.
68. **Hirtz D, Thurman DJ, Gwinn-Hardy K, et al (2007).** "How common are the "common" neurologic disorders?". *Neurology* **68** (5): 326–37.
69. **Illicki A, Larsson A(1988).** Psychomotor development of children with congenital hypothyroidism diagnosed by neonatal screening. *Acta Paediatr Scand* . 77: 142–147.
70. **Illingworth RS (1978).** Child development. In: Forfar JO, Arneil GC, eds. *Textbook of pediatrics.* Edinburgh, Scotland: Churchill Livingstone.
71. **James William D.; Berger, Timothy G.; et al. (2006).** *Andrews' Diseases of the Skin: clinical Dermatology.* Saunders Elsevier.
72. **Jiang Y-H, Lev-Lehman E, Bressler J, et al (1999)** Tsai TF, Beaudet AL. Genetics of Angelman syndrome. *Am J Hum Genet.* 65 :1 –6.
73. **Johnson JH, Goldman J (1990).** *Developmental assessment in clinical child psychology.* New York: Pergamon Press .
74. **Johnson LH, Bhutani VK, Brown AK(2002).** System-based approach to management of neonatal jaundice and prevention of kernicterus. *J Pediatr.* 140(4):396-403.
75. **Jones G, Steketee RW, Black RE, et al(2003).** How many child deaths can we prevent this year? *Lancet* 362:65–71.

76. **Karri, Sk; Saper, Rb; Kales, Sn (Jan 2008).** "Lead encephalopathy due to traditional medicines". *Current drug safety* 3 (1): 54–9.
77. **Knight SJ, Regan R, Nicod A, et al (1999).** Subtle chromosomal rearrangements in children with unexplained mental retardation. *Lancet.* 354 :1676 –1681.
78. **Kodo N, Millington DS, Norwood DL, et al (1992).** Automated analysis of free and short-chain acylcarnitine in plasma with a centrifugal analyzer. *Clin Chem* 38:2215-2221.
79. **Kolawole TM, Patel PJ, Mahdi AH (1989).** "Computed tomographic (CT) scans in cerebral palsy (CP)". *Pediatr Radiol* 20 (1–2): 23–7.
80. **Konstantareas MM, Beitchman JH(1991),** eds. Pervasive developmental disorders. *Psychiatr Clin North Am* 14:1-217
81. **Kosnett, M.J. (2007).** "Heavy metal intoxication and chelators". In Katzung, B.G.. *Basic and Clinical Pharmacology*. McGraw-Hill Professional.
82. **Krauss MJ, Morrissey AE, Winn HN, et al (2003).** *Am J Obstet Gynecol* 188:1484-90.
83. **Kravetz JD, Federman DG(2005);** Toxoplasmosis in pregnancy. *Am J Med.* 118(3):212-6.
84. **Kwok SK, Ho PC, Chan AK, et al (1996)** defects in children and adolescents with severe mental deficiency. *J Intellect Disabil Res* . 40: 330–335.
85. **Kyllerman, M.; Skjeldal, O. H.; Lundberg, et al (1994) :** Dystonia and dyskinesia in glutaric aciduria type I: clinical heterogeneity and therapeutic considerations. *Mov. Disord.* 9: 22-30,

86. **Laumonnier F, Bonnet-Brilhout F, Gomot M, et al (2004).** X-linked mental retardation and autism are associated with a mutation in the NLGN4 gene, a member of the neuroligin family. *Am J Med Genet.* 74 :552 –557.
87. **Levy SE, Hyman SL (1993).** Pediatric assessment of the child with developmental delay. *Pediatr Clin North Am* . 1993; 40: 465–477.
88. **Lewis R. First, and Judith S. Palfrey(1994).** The Infant or Young Child with Developmental Delay, Volume 330:478-483.
89. **Lingam S, Read S, Holland IM, et al (1982)** Wilson J, Brett EM, Hoare RD. Value of computerised tomography in children with non-specific mental subnormality. *Arch Dis Child.* 57 :381 –38.
90. **Lubs HA (1969).** "A marker X chromosome". *Am. J. Hum. Genet.* 21 (3): 231– 233.
91. **Luckasson RL, Coulter DL, Polloway EA, et al (1992).** Mental retardation: definition, classification and systems of supports. Washington: AAMR.
92. **MacLennan A(1999).** A template for defining a causal relation between acute intrapartum events and cerebral palsy: international consensus statement. *BMJ* 319:1054–1059.
93. **Mahfoud A, Dominguez CL, Rizzo C, Ribes A(2003).** "Rhabdomyolysis in glutaric aciduria type I.". *Rev Neurol* 39 (10): 939–42.
94. **Majnemer A (1998).** Benefits of early intervention for children with developmental disabilities. *Semin Pediatr Neurol* . 1998; 5: 62–69

95. **Majnemer A, Rosenbaum P, et al(2001)** Etiologic yield of autistic spectrum disorders: a prospective study. *J Child Neurol.* 16 :509–512.
96. **Majnemer A, Shevell MI (1995).** Diagnostic yield of the neurologic assessment of the developmentally delayed child. *J Pediatr* . 127: 193–199
97. **Malm G, Engman ML(2007);** Congenital cytomegalovirus infections. *Semin Fetal Neonatal Med.* 12(3):154-9.
98. **Martin, J. P. & Bell, J (1943).** "A pedigree of mental defect showing sex-linkage". *Journal of neurology, neurosurgery, and psychiatry (J. Neurol. Psychiat.)*. BMJ Publishing Group, London 6. , 154-157.
99. **Matsui, Sm; Mahoney, Mj; Rosenberg, Le (1983).** "The natural history of the inherited methylmalonic acidemias" . *The New England journal of medicine* 308 (15): 857–61.
100. **Mehta NM, Thomas RM(2002);** Antenatal screening for rubella-infection or immunity?; *BMJ.* 13;325(7355):90-1.
101. **Meisels SJ, Provence S.(1992)** Screening and assessment: guidelines for identifying young disabled and developmentally vulnerable children and their families. Washington, D.C.: National Center for Clinical Infant Programs.
102. **Menacker SJ(1993).** Visual function in children with developmental disabilities. *Pediatr Clin North Am* . 40: 659–674.
103. **Menyuk P(1986).** Predicting speech and language problems with persistent otitis media. In: Kavanagh JF, ed. *Otitis media and child development*. Parkton, Md.: York Press, 83-96.
104. **Milhorat TH, Bolognese PA, Nishikawa M, et al (2007).** "Syndrome of occipitoatlantoaxial hypermobility, cranial

- settling, and chiari malformation type I in patients with hereditary disorders of connective tissue". Journal of Neurosurgery: Spine 7 (6): 601–9.
105. **Minussi L, Mohrdieck R, Bercini M, et al(2008);** Prospective evaluation of pregnant women vaccinated against rubella in southern Brazil. *Reprod Toxicol.* 25(1):120-3.
106. **Moeschler John B, Michael Shevell(2006),** Clinical Genetic Evaluation of the Child With Mental Retardation or Developmental Delays, *PEDIATRICS* Vol. 117 No.pp. 2304-2316.
107. **Morton DH, Robinson DL, Puffenberger EG, et al (2003).** "Type I glutaric aciduria, part 1: natural history of 77 patients" . *Am J Med Genet C Semin Med Genet* **121** (1): 38–52.
108. **Msall ME, Bier JA, LaGasse L,(1998)** Tremont M, Lester B. The vulnerable preschool child: the impact of biomedical and social risks on neurodevelopmental function. *Semin Pediatr Neurol* . 1998; 5: 52–61.
109. **Nayae RL, Peters EC, Bartholomew M, et al (1989).** Origins of cerebral palsy. *Am J Dis Child* 143:1154–1161.
110. **Needleman HL, ed. Human (1992)** lead exposure. Boca Raton: CRC Press.
111. **Nicholas G. Guerina, Ho-Wen Hsu, et al (1994)** and the New England Regional Toxoplasma Working Group *N Engl J Med* ; 330:1858-1863.
112. **Niswander K, Henson G, Elbourne D, et al (1984).** Adverse outcome of pregnancy and the quality of obstetric care. *Lancet* ;ii:827–31.
113. **Ogier de Baulny H, Saudubray JM (2002).** "Branched-chain organic acidurias". *Semin Neonatol.* 7 (1): 65–74.

114. **Oswyn G, Vince JD, Friesen H(2000).** Perinatal asphyxia at Port Moresby General Hospital: a study of incidence, risk factors and outcome. *P N G Med J* 43:110–120.
115. **Ozmen M.,Tatli B. and Aydinli N. et al (2005)** Etiologic evaluation in 247 children with global developmental delay at Istanbul Turkey, *J Trop Pediatr* 51 pp. 310–313.
116. **Palfrey JS (1992).** Legislation for the education of children with disabilities. In: Levine MP, Carey WB, Crocker AC, eds. *Developmental-behavioral pediatrics*. 2nd ed. Philadelphia: W.B. Saunders, 782-5.
117. **Pharoah PO (2005).** "Causal hypothesis for some congenital anomalies". *Twin Res Hum Genet* 8 (6): 543–50.
118. **Phillips S, Sarles RM, Friedman SB(1992).** Consultation and referral for behavioral and developmental problems. In: Hoekelman RA, Friedman SB, Nelson NM, Seidel HM, eds. *Primary pediatric care*. 2nd ed. St. Louis: Mosby-Year Book, 678-82.
119. **Pollitt RJ, Green A, McCabe CJ, et al(1997).** Neonatal screening for inborn errors of metabolism: cost, yield and outcome. *Health Technol Assess* . 1: i-iv.
120. **Rapin I(1991).** Autistic children: diagnosis and clinical features. *Pediatrics* 87:751-760.
121. **Reinecke CJ, Mienie LJ, Hitzeroth HW, et al (1983).** Screening for inborn error of metabolism among mentally retarded patients. Outcome of a survey at the Witrand Care and Rehabilitation Center. *S Afr Med J* . 63: 14–16.

122. **Roschinger W, Olgemoller B, Fingerhut R, et al (2003)**
Advances in analytical mass spectrometry to improve screening for inherited metabolic diseases. *Eur J Pediatr.* 162 (suppl 1):S67–S76.
123. **Rosenbaum, RB; DP Ciaverella (2004).** Neurology in Clinical Practice. Butterworth Heinemann. pp. 2192–2193.
124. **Rosenberg MJ, Killoran C, Dziadzio L, et al (2001).**
Scanning for telomeric deletions **and** duplications and uniparental disomy using genetic markers in 120 children with malformations. *Hum Genet.* 2001;109 :311 –318.
125. **Rupa V(1995).** Dilemmas in auditory assessment of developmentally retarded children using behavioural observation audiometry and brain stem evoked response audiometry. *J Laryngol Otol* . 109: 605–609.
126. **Sáez-Llorens X, McCracken GH (June 2003).** "Bacterial meningitis in children". *Lancet* 361 (9375): 2139–48.
127. **Saigal S, Szatmari P, Rosenbaum P, et al (1990).**
Intellectual and functional status at school entry of children who weighed 1000 grams or less at birth: a regional perspective of births in the 1980's. *J Pediatr* 116:409-416.
128. **Saudubray J, Sedel F, Walter J (2006).** "Clinical approach to treatable inborn metabolic diseases: an introduction". *J Inherit Metab Dis* 29 (2-3): 261–74.
129. **Schaefer GB, Bodensteiner JB (1992).** Evaluation of the child with idiopathic mental retardation. *Pediatr Clin North Am* . 1992; 39: 929–943.
130. **Schaefer GB, Bodensteiner JB(1998).** Radiological findings in developmental delay. *Semin Pediatr Neurol.* 36 :33 – 38.

131. **Schaefer GB, Bodensteiner JB(1998).** Radiological findings in developmental delay. *Semin Pediatr Neurol.* 36 :33 –38
132. **Schmitt B, Seeger J, Kreuz E, et al (1991).** Central nervous system involvement of children with HIV infection. *Dev Med Child Neurol* 33:535-540.
133. **Shaag, A.; Saada, A ; Berger, I,et al (1999) :** Molecular basis of lipoamide dehydrogenase deficiency in Ashkenazi Jews. *Am. J. Med. Genet.*82:177-182.
134. **Shalowitz MU, Gorski PA (1990).** Developmental assessment in the first year of life. In: Stockman JA III, ed. *Difficult diagnosis in pediatrics.* Philadelphia: W.B. Saunders, 3-14.
135. **Sherman, S. (2002).** "Epidemiology". Chapter 3 in *Fragile X Syndrome, Diagnosis Treatment and Research.* Ed. Hagerman, R. J. & Hagerman, P.J. (3rd Edition). Johns Hopkins University Press: Baltimore.
136. **Shevell M MD, CM, FRCPC (2008).** Global Developmental Delay and Mental Retardation or Intellectual Disability: Conceptualization, Evaluation, and Etiology *Pediatric Clinics of North America* Volume 55, Issue 5, Developmental Disabilities, Part I Pages 1071-1084.
137. **Shevell M, Ashwal S, Donley D, et al (2003).** Practice parameter: Evaluation of the child with global developmental delay: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. *Neurology.* 60 :367 –380.
138. **Shevell M.I. and Sherr E. (2006),** Global developmental delay and mental retardation. In: K. Swaiman, S. Ashwal and D.

- Ferreiro, Editors, *Pediatric Neurology: Principles & Practice* ((ed 4).), Mosby Elsevier, Philadelphia, PA pp. 799–820.
139. **Shevell M. MD, CM, FRCPC (2006).** Office evaluation of the child with developmental delay, *Practical Office Management of Common Pediatric Neurology Problems Volume 13, Issue 4, Pages 256-261,*
 140. **Shevell M.I., Majnemer A. and Morin I.,(2003)** Etiologic yield of cerebral palsy: A contemporary case-series, *Pediatr Neurol* **28** , pp. 352–359.
 141. **Shevell M.I., Majnemer A. and Rosenbaum P. et al.,(2001)** Etiologic yield of autistic spectrum disorders: A prospective study, *J Child Neurol* **16** (2001), pp. 509–512.
 142. **Shevell M.I., Ashwal S. and Donley D. et al.,(2003)** Practice parameter: Evaluation of the child with global developmental delay, *Neurology* **60** (2003), pp. 367–379.
 143. **Shevell M.I., Majnemer A. and Rosenbaum P. et al.,(2000)** Etiologic yield in single domain developmental delay: A prospective study, *J Pediatr* **137** (2000), pp. 633–637.
 144. **Shevell MI (1998).** The evaluation of the child with a global developmental delay. *Semin Pediatr Neurol* . 1998; 5: 21–25.
 145. **Shevell MI, Majnemer A, Rosenbaum P, Abrahamowicz M(2001).** Etiologic yield of autistic spectrum disorders: a prospective study. *J Child Neurol.* 16 :509 –512.
 146. **Shevell MI, Majnemer A, Rosenbaum P, et al (2000)** Etiologic yield of subspecialists' evaluation of young children with global developmental delay. *J Pediatr.* 136 :593 –598.
 147. **Shevell MI, Majnemer A, Rosenbaum P, et al (2001)** Etiologic determination of childhood developmental delay. *Brain*

- Dev. 2001;23 :228 –235.
148. **Shonkoff JP, Hauser-Cram P(1987).** Early intervention for disabled infants and their families: a quantitative analysis. *Pediatrics* 80:650-658.
 149. **Simeonsson RJ, Sharp MC (1992).** Developmental delays. In: Hoekelman RA, Friedman SB, Nelson NM, et al., eds. *Primary pediatric care*. St. Louis: Mosby-Year Book, 867–870.
 150. **Slavotinek A, Rosenberg M, Knight S, et al (1999).** Screening for submicroscopic chromosome rearrangements in children with idiopathic mental retardation using microsatellite markers for the chromosome telomeres. *J Med Genet.* 36 :405 – 411.
 151. **Srour M., Mazer B. and M.I. Shevell M.I., (2006)** Analysis of clinical features predicting etiologic yield in the assessment of global developmental delay, *Pediatrics* **118** , pp. 139–145.
 152. **Straus SE, Thorpe KE, Holroyd-Leduc J (2006).** "How do I perform a lumbar puncture and analyze the results to diagnose bacterial meningitis?". *JAMA : the journal of the American Medical Association* **296** (16): 2012–22.
 153. **Szabó, Nóra; Pap, Csenge; Kóbor, Jenő; et al (2010).** LászlóActa Pædiatrica, Volume 99, Number 5, pp. 690-693.
 154. **Tarim OF, Yordam N(1992).** Congenital hypothyroidism in Turkey: a retrospective evaluation of 1000 cases. *Turk J Pediatr* . 34: 197–202.
 155. **Thompson MGG(1990).** Developmental assessment of the preschool child. In: Stockman JA III, ed. *Difficult diagnosis in pediatrics*. Philadelphia: W.B. Saunders, 15-28.

156. **Thorngren-Jerneck K, Herbst A (2006).** "Perinatal factors associated with cerebral palsy in children born in Sweden". *Obstetrics and gynecology* **108** (6): 1499–505.
157. **Timbrell, J.A., ed (2008).** "Biochemical mechanisms of toxicity: Specific examples". *Principles of Biochemical Toxicology*, 4th edition. Informa Health Care.
158. **Tjio J.H & Levan A (1956).** The chromosome number of man. *Hereditas* 42, 1-6. .
159. **Tortorelli, S.; Hahn, S. H.; Cowan, et al (2005):** The urinary excretion of glutaryl carnitine is an informative tool in the biochemical diagnosis of glutaric acidemia type I. *Molec. Genet. Metab.* 84: 137-143.
160. **Tunkel AR, Hartman BJ, Kaplan SL (2004).** "Practice guidelines for the management of bacterial meningitis". *Clinical Infectious Diseases* 39 (9): 1267–84.
161. **Van Karnebeek CD, Jansweijer MC, Leenders AG, et al (2005).** Diagnostic investigations in individuals with mental retardation: a systematic literature review of their usefulness. *Eur J Hum Genet.* 13 :6 –25.
162. **van Karnebeek CD, Jansweijer MC, Leenders AG, et al (2005).** Diagnostic investigations in individuals with mental retardation: a systematic literature review of their usefulness. *Eur J Hum Genet.* 13 :6 –25.
163. **Van Karnebeek CD, Koevoets C, Sluijter S, et al (2002).** Prospective screening for subtelomeric rearrangements in children with mental retardation of unknown aetiology: the Amsterdam experience. *J Med Genet.* 39 :546 –553.

164. **Van Karnebeek CDM, Scheper FY, Abeling NG, et al (2002).** Amsterdam, Netherlands: Department of Pediatrics/Emma Children's Hospital and Department of Clinical Genetics, Academic Medical Centre, University of Amsterdam; 75–108.
165. **Walker D, Gugenheim S, Downs MP, et al (1989).** Early Language Milestone Scale and language screening of young children. *Pediatrics* 83:284-288.
166. **Warburg M(1994).** Visual impairment among people with developmental delay. *J Intellect Disabil Res* . 38: 423–432.
167. **Wendy S Meschino, MD FRCPC FCCMG (2003).** Department of Genetics, North York General Hospital, Toronto, OntarioCorrespondence: Dr Wendy S Meschino, Clinical Geneticist, Department of Genetics, North York General Hospital.
168. **White BJ, Ayad M, Fraser A, et al (1999).** A 6-year experience demonstrates the utility of screening for both cytogenetic and FMR-1 abnormalities in patients with mental retardation. *Genet Test* . 3: 291–296.
169. **Willi SM, Moshang Jr T(1991).** Results of screening tests for congenital hypothyroidism. *Pediatr Clin North Am* . 38: 555–566.
170. **Yaqub BA(1996).** Subacute sclerosing panencephalities (SSPE): early diagnosis, prognostic factors and natural history. *J Neurol Sci* 139: 227–234.
171. **Yeargin-Allsopp M, Murphy CC, Cordero JF, et al (1997).** Reported biomedical causes and associated medical conditions for mental retardation among 10 year old children, metropolitan Atlanta, 1985 to 1987. *Dev Med Child Neurol* . 39: 142–149.

172. **Yeum CH, Kim SW, Kim NH, Choi KC, Lee J (2002).**
 "Increased expression of aquaporin water channels in hypothyroid
 rat kidney". *Pharmacol. Res.* **46** (1): 85–88.
173. **Zarmeneh Aly, Fawad Taj, and Shahnaz Ibrahim**
 (2010). *brain and development* Volume 32, Issue 2, Pages 90-97
174. **Zoghbi HY (2003).** Postnatal neurodevelopmental disorders:
 meeting at the synapse? *Science.* 302 :826 –830.
175. **Zuckerman B, Bresnahan K (1991).** Developmental and
 behavioral consequences of prenatal drug and alcohol exposure.
 Pediatr Clin North Am 38:1387-1406.