

References

- Aimaretti G, Corneli G and Razzore P (2002) :

Comparison between insulin-induced hypoglycemia and growth hormone (GH)-releasing hormone + arginine as provocative tests for the diagnosis of GH deficiency in adults . J. Clin. Endocrinol. Metab. 83 (5): 1615–8.

- Albertson R, Riggs B and Sullivan W (2005):

Membrane traffic: a driving force in cytokinesis . Trends Cell Biol 15 (2): 92–101.

- Alfirevic Z, Sundberg K and Brigham S (2003) :

Amniocentesis and chorionic villus sampling for prenatal diagnosis. Cochrane Database Syst Rev.:(3):CD003252.

-Allison; Ross,PL Beales and J Hill (2008) :

The Clinical, Molecular, and Functional Genetics of Bardet-Biedl Syndrome, in Genetics of Obesity Syndromes. Oxford University Press. pp. 153-4

- Andy C, Flake D and French L (2005) :

Clinical inquiries. Do insulin-sensitizing drugs increase ovulation rates for women with PCOS? . J Fam Pract 54 (2): 159–60.

- Ansley SJ, Badano JL, Blacque OE, Hill J, Hoskins BE, Leitch CC, Kim JC, Ross AJ, Eichers ER, Teslovich TM, Mah AK, Johnsen RC, Cavender JC, Lewis RA, Leroux MR, Beales PL and Katsanis N (2003) :

Basal body dysfunction is a likely cause of pleiotropic Bardet-Biedl syndrome . Nature 425 (6958): 628–33.

- Antonarakis, S. E (2004) :

Chromosome 21 and Down syndrome: From genomics to pathophysiology. Nature Reviews Genetics 5, 725–38 .

- **Arlan Rosenbloom, Janet H and Silverstein (2003) :**

Type 2 Diabetes in Children and Adolescents: A Clinician's Guide to Diagnosis, Epidemiology, Pathogenesis, Prevention, and Treatment. American Diabetes Association, U.S. pp. 1.

- **Armand Marie Leroi (2003) :**

Mutants: on the form, varieties and errors of the human body. London: HarperCollins. pp. 16–17.

- **Azzari C, Gambineri E, Moriondo M, Betti L and Saldias LR (2005) :**

Safety and immunogenicity of measles-mumps-rubella vaccine in children with congenital immunodeficiency (DiGeorge syndrome). Vaccine. ;23(14):1668-71.

- **Azaiez H, Yang T and Prasad S (2007) :**

Genotype-phenotype correlations for SLC26A4-related deafness . Hum. Genet. 122 (5): 451–7.

- **Azziz R (2004 Jun) :**

The prevalence and features of the polycystic ovary syndrome in an unselected population. J Clin Endocrinol Metab.;89(6):2745-9.

- **Badano JL, Mitsuma N, Beales PL and Katsanis N (2006) :**

The ciliopathies: an emerging class of human genetic disorders. Annu Rev Genomics Hum Genet 7: 125–48.

- **Bardoni B, Mandel JL and Fisch GS (2000) :**

FMR1 gene and fragile X syndrome. Am J Med Genet. ;97(2):153-63.

- **Beales P, Elcioglu N, Woolf A, Parker D and Flintner F (2000) :**

New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey . J. Med. Genet. 36 (6): 437–46.

- Beam D, Poe MD, Provenzale JM, Szabolcs P, Martin PL, Prasad V, Parikh S, Driscoll T, Mukundan S, Kurtzberg J and Escolar ML (2007) :

Outcomes of unrelated umbilical cord blood transplantation for X-linked adrenoleukodystrophy. Biol Blood Marrow Transplant 13 (6): 665–74.

- Bejerano G, Lowe CB, Ahituv N, King B and Siepel A (2006) :

A distal enhancer and an ultraconserved exon are derived from a novel retroposon. Nature 441: 87–90.

- Bianco B, Lipay MV, Melaragno MI, Guedes AD and Verreschi IT (Sep 2006) :

Detection of hidden Y mosaicism in Turner's syndrome: importance in the prevention of gonadoblastoma. J Pediatr Endocrinol Metab. ;19(9):1113-7.

- Biemont C and Vieira C (2006) :

Genetics—Junk DNA as an evolutionary force. Nature 443: 521–4

-Blake MA, Kalra MK and Maher MM (2004) :

Pheochromocytoma: an imaging chameleon. Radiographics;24 Suppl 1:S 87-99.

- Bolar K, Hoffman AR, Maneatis T and Lippe B (2008) :

Long-term safety of recombinant human growth hormone in Turner syndrome . J. Clin. Endocrinol. Metab. 93 (2): 344–51.

- Bondy CA (Jan 2007) :

Care of girls and women with Turner syndrome: A guideline of the Turner Syndrome Study Group. J Clin Endocrinol Metab. ;92(1):10-25.

- Borck . G, A. K. Topaloglu, E. Korsch, U. Martine, G. Wildhardt, N. Onenli-Mungan, B. Yuksel, U. Aumann, G. Koch and G. Ozer (2005) :

Four New Cases of Congenital Secondary Hypothyroidism due to a Splice Site

Mutation in the Thyrotropin- β Gene: Phenotypic Variability and Founder Effect . J. Clin. Endocrinol. Metab., 189(8): 4136 –41

-Braig M and Schmitt C (2006) :

Oncogene-induced senescence: putting the brakes on tumor development .
Cancer Res 66 (6): 2881–4.

- Brambati B and Tului L (2005 Apr) :

Chorionic villus sampling and amniocentesis. Curr Opin Obstet
Gynecol.;17(2):197-201.

- Bringham FR, Demay MB and Kronenberg HM (2008) :

Disorders of Mineral Metabolism. In: Kronenberg HM, Schlomo M, Polansky
KS, Larsen PR, eds. Williams Textbook of Endocrinology. 11th ed. St. Louis,
Mo: WB Saunders .chap. 27.

- Byrne J (Jul 1999) :

Infertility and premature menopause in childhood cancer survivors. Med Pediatr
Oncol. ;33(1):24-8.

-Callender GG, Rich TA and Perrier ND (Aug 2008) :

Multiple endocrine neoplasia syndromes . Surg. Clin. North Am. 88 (4): 863–
95

**- Camastra S, Bonora E, Del Prato S, Rett K, Weck M and Ferrannini E
(December 1999) :**

Effect of obesity and insulin resistance on resting and glucose-induced
thermogenesis in man. EGIR (European Group for the Study of Insulin
Resistance) . Int. J. Obes. Relat. Metab. Disord. 23 (12): 1307–13.

- **Carlo Gelmetti , Caputo and Ruggero (2002) :**

Pediatric Dermatology and Dermatopathology: A Concise Atlas. T&F STM.
p. 160.

- **Carmina E (2004 Feb) :**

Diagnosis of polycystic ovary syndrome: from NIH criteria to ESHRE-ASRM guidelines. Minerva Ginecol.;56(1):1-6.

- **Carney JA (Feb 2005) :**

Familial multiple endocrine neoplasia: the first 100 years . Am. J. Surg. Pathol.
29 (2): 254–74.

- **Carninci P and Hayashizaki Y (2007) :**

Noncoding RNA transcription beyond annotated genes . Curr. Opin. Genet.
Dev. 17 (2): 139–44.

- **Carrel AL, Myers SE, Whitman BY and Allen DB (April 2002) :**

Benefits of long-term GH therapy in Prader-Willi syndrome: a 4-year study. J.
Clin. Endocrinol. Metab. 87 (4): 1581–5.

- **Carrel L and Willard H (2005) :**

X-inactivation profile reveals extensive variability in X-linked gene expression
in females. Nature 434 (7031): 400–4.

- **Caughey AB, Hopkins LM and Norton ME (2006 Sep) :**

Chorionic villus sampling compared with amniocentesis and the difference in
the rate of pregnancy loss. Obstet Gynecol.;108(3 Pt 1):612-6.

- **Chace DH, Kalas TA and Naylor EW (2003) :**

Use of tandem mass spectrometry for multianalyte screening of dried blood
specimens from newborns. Clin. Chem. 49 (11): 1797–817.

- **Chang AS, Moley KH, Wangler M, Feinberg AP and Debaun MR (February 2005) :**

Association between Beckwith-Wiedemann syndrome and assisted reproductive technology: a case series of 19 patients. *Fertility and sterility* 83 (2): 349–54.

- **Chan G, Liu S and Yen T (2005) :**

"Kinetochore structure and function". *Trends Cell Biol* 15 (11): 589–98.

- **Charlesworth B (2003) :**

Sex Determination in the Honeybee. *Cell* 114 (4): 397–8.

- **Chau V, Karvelas G, Jacob P and Carmant L (2004) :**

Early treatment of Aicardi syndrome with vigabatrin can improve outcome. *Neurology*;63(9):1756-7.

- **Cheadle C, Fan J, Cho-Chung YS, Werner T, Ray J, Do L, Gorospe M and Becker KG (2005) :**

Control of gene expression during T cell activation: alternate regulation of mRNA transcription and mRNA stability. *BMC Genomics* 6 (1): 75.

- **Chen CJ and Fischbeck KH (2006) :**

Clinical aspects and the genetic and molecular biology of Kennedy's disease". in Tetsuo Ashizawa; Wells, Robert V. *Genetic Instabilities and Neurological Diseases* (Second Edition ed.). Boston: Academic Press Chapter 13. pp. 211–22.

- **Chen CP (May 1 2006) :**

Aplasia and duplication of the thumb and facial clefts associated with fetal trisomy 18. *Am J Med Genet A.* ;140(9):960-3.

- **Chitturi S, Harris M and Thomsett MJ (2008) :**

Utility of AVP gene testing in familial neurohypophyseal diabetes insipidus .
Clin. Endocrinol. (Oxf) 69 (6): 926–30 .

- **Chow J, Yen Z, Ziesche S and Brown C (2005) :**

Silencing of the mammalian X chromosome . Annu Rev Genomics Hum Genet
6: 69-92.

- **Cho YM, Kim M, Park KS, Kim SY and Lee HK (May 2003) :**

S20G mutation of the amylin gene is associated with a lower body mass index
in Korean type 2 diabetic patients. Diabetes Res. Clin. Pract. **60** (2): 125–9.

- **Clague A and Thomas A (2002) :**

Neonatal biochemical screening for disease. Clin. Chim. Acta 315 (1-2): 99–
110.

- **Clericuzio CL, Chen E and McNeil DE (August 2003) :**

Serum alpha-fetoprotein screening for hepatoblastoma in children with
Beckwith-Wiedemann syndrome or isolated hemihyperplasia . The Journal of
pediatrics 143 (2): 270–2.

- **Cohen BA, Mitra RD, Hughes JD and Church GM (2000) :**

A computational analysis of whole-genome expression data reveals
chromosomal domains of gene expression. Nature Genet 26: 183–6.

- **Concha Ruiz M (2006) :**

Surgical treatment of the aortic root dilatation (in Spanish; Castilian). An R
Acad Nac Med (Madr) 123 (3): 557–68; discussion 569–71.

- **Consugar MB, Anderson SA, Rossetti S, Pankratz VS, Ward CJ, Torra R,
Coto E, El-Youssef HM, Kantarci S, Utsch B, Hildebrandt F, Sweeney WE,
Avner ED, Torres VE, Cunningham JM and Harris PC (2005) :**

Haplotype analysis improves molecular diagnostics of autosomal recessive polycystic kidney disease. *Am J Kidney Dis* 45 : 77 –87,

- ***Costanzi C, Stein P, Worrall DM, Schultz RM and Pehrson JR (2000) :***

Histone macroH2A1 is concentrated in the inactive X chromosome of female preimplantation mouse embryos. *Development* 127: 2283–9.

- ***Cotran, Kumar and Collins (1999) :***

Robbins Pathologic Basis of Disease, Saunders Sixth Edition,; 913-26.

- ***Ristine Cortet-Rudelli and Didier Dewailly (Sep 21 2006) :***

Diagnosis of Hyperandrogenism in Female Adolescents. Hyperandrogenism in Adolescent Girls. Armenian Health Network, Health.am.

- ***Cutting GR (2005) :***

Modifier genetics: Cystic fibrosis. *Annu Rev Genomics Hum Genet* 6 : 237 – 60,

- ***Daly and Lewis Landsberg (2008):***

Multiple endocrine neoplasia syndrome ,Hormonal disorder Merck manual Pag :345-9

- ***Davenport ML, Crowe BJ and Travers SH (2007) :***

Growth hormone treatment of early growth failure in toddlers with Turner syndrome: a randomized, controlled, multicenter trial . *J Clin Endocrinol Metab.* 92 (9): 3406–16 .

- ***Dean L and McEntyre J (2004) :***

The Genetic Landscape of Diabetes. Bethesda:NCBI, 2004. This is an entire online textbook on the complex genetics of the forms of diabetes. The chapter on MODY provides an up-to-date and concise overview of the molecular

defects. Little expansion of clinical knowledge of the 6 types since 2001 has occurred.

- DeBaun MR, Niemitz EL, McNeil DE, Brandenburg SA, Lee MP and Feinberg AP (March 2002) :

Epigenetic alterations of H19 and LIT1 distinguish patients with Beckwith-Wiedemann syndrome with cancer and birth defects. American journal of human genetics 70 (3): 604–11

- Denschlag, Dominik, MD; Clemens, Tempfer, MD; Kunze, Myriam, MD; Wolff, Gerhard, MD; Keck and Christoph, MD (2004) :

Assisted reproductive techniques in patients with Klinefelter syndrome: A critical review, Fertility and Sterility 82 (4): 775–9 .

- Dequéant ML and Pourquié O (2008) :

Segmental patterning of the vertebrate embryonic axis. Nat Rev Genet.;9(5):370-82.

- De Smith AJ, Purmann C and Walters RG (June 2009) :

A Deletion of the HBII-85 Class of Small Nucleolar RNAs (snoRNAs) is Associated with Hyperphagia, Obesity and Hypogonadism. Hum. Mol. Genet.

- De Souza CP and Osmani SA (2007) :

Mitosis, not just open or closed. Eukaryotic Cell 6 (9): 1521–7.

- Ding F, Li HH, Zhang S, Solomon NM, Camper SA, Cohen P and Francke U (2008) :

SnoRNA Snord116 (Pwcr1/MBII-85) deletion causes growth deficiency and hyperphagia in mice.. PLoS ONE 3 (3): e1709.

- Ding F, Prints Y, Dhar MS, Johnson DK, Garnacho-Montero C, Nicholls RD and Francke U (2005) :

Lack of Pwcr1/MBII-85 snoRNA is critical for neonatal lethality in Prader-Willi syndrome mouse models.. Mamm Genome 16 (6): 424–31.

- **Donaldson MD, Gault EJ, Tan KW and Dunger DB (Jun 2006) :**

Optimising management in Turner syndrome: from infancy to adult transfer. Arch Dis Child. ;91(6):513-20.

- **Driscoll DA and Gross S (2005) :**

Prenatal screening for aneuploidy, N Engl J Med, 360:2556 - 58

- **Durai S, Mani M, Kandavelou K, Wu J, Porteus MH and Chandrasegaran S (2005) :**

Zinc finger nucleases: custom-designed molecular scissors for genome engineering of plant and mammalian cells. Nucleic Acids Res. 33 (18): 5978–90.

- **Dykens EM (2007) :**

Psychiatric and behavioral disorders in persons with Down syndrome. Ment Retard Dev Disabil Res Rev 13 (3): 272–8.

- **Eberhart, M. S.; Ogden, C, Engelgau, M, Cadwell, B, Hedley, A. A. and Saydah, S. H (November 2004) :**

Prevalence of Overweight and Obesity Among Adults with Diagnosed Diabetes --- United States, 1988--1994 and 1999--2002. Morbidity and Mortality Weekly Report (Centers for Disease Control and Prevention) 53 (45): 1066–8.

- **Eddleman, K.A (November 2006) :**

Pregnancy Loss Rates After Midtrimester Amniocentesis. Obstetrics and Gynecology, volume 108, number 5, pages 1067-72.

- **Edelmann L and Hirschhorn K (2009 Jan) :**

Clinical utility of array CGH for the detection of chromosomal imbalances

associated with mental retardation and multiple congenital anomalies. Ann N Y Acad Sci.;1151:157-66.

- *El-Hajj Fuleihan, G. and Vieth, R (2006) :*

Vitamin D insufficiency and musculoskeletal health in children and adolescents. Int. Congr. Ser. **1297**, 91–108.

-*Ellis NA, Ciocchi S and German J (2001) :*

Back mutation can produce phenotype reversion in Bloom syndrome somatic cells. Hum Gen135-

- *Engelen M, Ofman R and Dijkgraaf MG (January 2010) :*

Lovastatin in X-linked adrenoleukodystrophy. The New England Journal of Medicine 362 (3): 276–7.

-*Epstein CJ (2006) :*

Down's syndrome: critical genes in a critical region. Nature 441 (7093): 582–3.

- *Fajans SS, Bell GI and Polonsky KS (2001) :*

Molecular mechanisms and clinical pathophysiology of maturity-onset diabetes of the young . N. Engl. J. Med. **345** (13): 971–80.

- *Fernandez L, Lapunzina P and Arjona D (Oct 2005) :*

Comparative study of three diagnostic approaches (FISH, STRs and MLPA) in 30 patients with 22q11.2 deletion syndrome. Clin Genet. ;68(4):373-8.

- *Finsterer J (2007) :*

Hematological manifestations of primary mitochondrial disorders. Acta Haematol. 118 (2): 88–98.

- *Fischbeck KH, Lieberman A, Bailey CK, Abel A and Merry DE (2000) :*

Androgen receptor mutation in Kennedy's disease . Philos. Trans. R. Soc. Lond., B, Biol. Sci. 354 (1386): 1075–8.

- ***Fiumara A, Sorge G and Toscano A (Jul 30 2004) :***

Perrault syndrome: evidence for progressive nervous system involvement. Am J Med Genet. ;128A(3):246-9.

- ***Fluck CE, Tajima T and Pandey AV (Mar 2004) :***

Mutant P450 oxidoreductase causes disordered steroidogenesis with and without Antley-Bixler syndrome. Nat Genet. ;36(3):228-30.

- ***Fogu G, Maserati E, Cambosu F, Moro MA, Poddie F and Soro G (2008) :***

Patau syndrome with long survival in a case of unusual mosaic trisomy 13. Eur J Med Genet. ;51(4):303-14.

- ***Galvani A and Slatkin M (2003) :***

Evaluating plague and smallpox as historical selective pressures for the CCR5- Δ 32 HIV-resistance allele . Proc Natl Acad Sci USA 100 (25): 15276–9

- ***Garguillo AR and Hill JA (1999) :***

Autoimmune endocrinopathies in female reproductive dysfunction. In: Volpe R, ed. Contemporary Endocrinology: Autoimmune Endocrinopathies. Totowa, NJ: Humana Press :365-91.

- ***Gartler SM, Varadarajan KR, Luo P, Canfield TK, Traynor J, Francke U and Hansen RS (2004) :***

Normal histone modifications on the inactive X chromosome in ICF and Rett syndrome cells: implications for methyl-CpG binding proteins . BMC Biology 2: 21.

- ***Gehring, W. J (2001) :***

The genetic control of eye development and its implications for the evolution of various eye-types . Zoology 104 (3–4): 171–81.

-Genuth S (2006) :

Insights from the diabetes control and complications trial/epidemiology of diabetes interventions and complications study on the use of intensive glycemic treatment to reduce the risk of complications of type 1 diabetes. *Endocr Pract* 12 (Suppl 1): 34–9

- Gerton, J. L. and Hawley, R. S (2005) :

Homologous chromosome interactions in meiosis: Diversity amidst conservation. *Nature Reviews Genetics* 6, 477–87

- Gicquel C, Gaston V, Mandelbaum J, Siffroi JP, Flahault A and Le Bouc Y (May 2003) :

In vitro fertilization may increase the risk of Beckwith-Wiedemann syndrome related to the abnormal imprinting of the KCN1OT gene . *American journal of human genetics* 72 (5): 1338–41.

- Gilbert SF (2003) :

Developmental biology, 7th ed., Sunderland, Mass: Sinauer Associates, 65–6.

- Glotzer M (2005) :

The molecular requirements for cytokinesis. *Science* 307 (5716): 1735–9.

- González F, Rote N, Minium J and Kirwan J (2006) :

Reactive oxygen species-induced oxidative stress in the development of insulin resistance and hyperandrogenism in polycystic ovary syndrome . *J Clin Endocrinol Metab* 91 (1): 336–40.

- Gosden R, Trasler J, Lucifero D and Faddy M (2003) :

Rare congenital disorders, imprinted genes, and assisted reproductive technology . *Lancet* **361** (9373): 1975–7.

- **Green-Golan L, Yates C and Drinkard B (Aug 2007) :**

Patients with classic congenital adrenal hyperplasia have decreased epinephrine reserve and defective glycemic control during prolonged moderate-intensity exercise. J Clin Endocrinol Metab. ;92(8):3019-24.

- **Grosso S, Pucci L, Di Bartolo RM, Gobbi G, Bartalini G, Anichini C, Scarinci R, Balestri M, Farnetani MA, Cioni M, Morgese G and Balestri P (2005) :** Chromosome 18 aberrations and epilepsy: a review. Am J Med Genet A;134:88–9.

- **Hagerman RJ, Leavitt BR and Farzin F (May 2004) :**

Fragile-X-associated tremor/ataxia syndrome (FXTAS) in females with the FMR1 premutation. Am J Hum Genet. ;74(5):1051-6.

-**Hagop Youssoufian, Reed E and Pyeritz (October 2002) :**

And consequences of somatic mosaicism in humans Human genetics and disease: Mechanisms . Nature Reviews Genetics 3, 748-58

- **Halliday J, Oke K, Breheny S, Algar E and J Amor D (September 2004) :**

Beckwith-Wiedemann syndrome and IVF: a case-control study . American journal of human genetics 75 (3): 526–8.

- **Haper,P (2004) :**

parictical genetic counseling ,London ,Arnold text ED.7th ch. 8 ,pag 476-83 .

- **Hardiman P, Pillay OC and Atiomo W (2003 May 24) :**

Polycystic ovary syndrome and endometrial carcinoma.Lancet. 361(9371):1810-2.

- **H J Lee, I H Park, H J Kim and S U Kim (2009) :**

Human neural stem cells overexpressing glial cell line-derived neurotrophic factor in experimental cerebral hemorrhage Gene Ther 16: 1066-76

- **Hoki Y, Kimura N, Kanbayashi M, Amakawa Y, Ohhata T, Sasaki H and Sado T (2009) :**

A proximal conserved repeat in the Xist gene is essential as a genomic element for X-inactivation in mouse. (abstract). Development 136: 139–46.

- **Höybye C, Hilding A, Jacobsson H and Thorén M (May 2003) :**

Growth hormone treatment improves body composition in adults with Prader-Willi syndrome.

- **Hu FB (February 2003) :**

Sedentary lifestyle and risk of obesity and type 2 diabetes. Lipids 38 (2): 103–8.

- **James, William D.; Berger and Timothy G (2006) :**

Andrews' Diseases of the Skin: clinical Dermatology. Saunders Elsevier. ISBN 0-7216-2921-0.

- **Jannink, J; Bink, Mc and Jansen (Aug 2001) :**

Using complex plant pedigrees to map valuable genes. Trends in plant science 6 (8): 337–42.

- **Jiménez C, Cote G, Arnold A and Gagel RF (2006) :**

Review: Should patients with apparently sporadic pheochromocytomas or paragangliomas be screened for hereditary syndromes?. J Clin Endocrinol Metab. ;91(8):2851-8.

- **Johnston NJ and Franklin DL (Mar 2006) :**

Dental findings of a child with Wolf-Hirschhorn syndrome. Int J Paediatr Dent. ;16(2):139-42.

- **Jorge Sequeiros and Bárbara Guimarães (2008) :**

Definitions of Genetic Testing , EuroGentest Network of Excellence Project .

- **Kagami, M.; Nishimura, G.; Okuyama, T.; Hayashidani, M.; Takeuchi, T.; Tanaka, S.; Ishino, F.; Kurosawa, K. and Ogata, T (2005):**

Segmental and full paternal isodisomy for chromosome 14 in three patients: Narrowing the critical region and implication for the clinical features. Am. J. Med. Genet. 138A: 127-32.

- **Kalantaridou SN, Calis KA, Vanderhoof VH, Bakalov VK, Corrigan EC and Troendle JF (Nov 2006) :**

Testosterone deficiency in young women with 46,XX spontaneous premature ovarian failure. Fertil Steril. ;86(5):1475-82.

- **Kalantaridou SN, Braddock DT, Patronas NJ and Nelson LM (Jul 1999):**

Treatment of autoimmune premature ovarian failure. Hum Reprod. ;14(7):1777-82.

- **Kalantaridou SN, Naka KK and Papanikolaou E (Aug 2004) :**

Impaired endothelial function in young women with premature ovarian failure: normalization with hormone therapy. J Clin Endocrinol Metab. ;89(8):3907-13.

- **Kalelioglu I, Kubat Uzun A, Yildirim A, Ozkan T, Gungor F and Has R (2007) :**

Transient gestational diabetes insipidus diagnosed in successive pregnancies: review of pathophysiology, diagnosis, treatment, and management of delivery. Pituitary 10 (1): 87–93. Clin. Endocrinol. (Oxf) 58 (5): 653–61.

- **Kantaputra PN, Limwongse C, Tochareontanaphol C, Mutirangura A, Mevatee U and Praphanphoj V (2006) :**

Contiguous gene syndrome of holoprosencephaly and hypotrichosis simplex: association with an 18p11.3 deletion. Am J Med Genet A.;140:2598–602.

- **Kayton A (2007) :**

Newborn screening: a literature review. Neonatal network : NN 26 (2): 85–95.

- **Kelly M, Robinson BW and Moore JW (Nov 1 2002) :**

Trisomy 18 in a 20-year-old woman. Am J Med Genet. ;112(4):397-9.

-**Kelman LM and Kelman Z (2004) :**

Multiple origins of replication in archaea . Trends Microbiol. 12 (9): 399–401.

- **Killeen and Anthony A (2004) :**

Genetic Inheritance. Principles of Molecular Pathology. Humana Press. pp. 41.

- **Kingsbury MA, Friedman B and McConnell MJ (2005) :**

Aneuploid neurons are functionally active and integrated into brain circuitry .

Proceedings of the National Academy of Sciences of the United States of

America 102 (17): 6143–7

- **Kirchlechner V, Koller DY, Seidl R and Waldhauser F (2007) :**

Treatment of nephrogenic diabetes insipidus with hydrochlorothiazide and
amiloride . Arch. Dis. Child. 80 (6): 548–52.

- **Knowler W, Barrett-Connor E, Fowler S, Hamman R, Lachin J, Walker E
and Nathan D (2002) :**

Reduction in the incidence of type 2 diabetes with lifestyle intervention or
metformin . N Engl J Med 346 (6): 393–403

- **Kobrynski LJ and Sullivan KE (Oct 20 2007) :**

Velocardiofacial syndrome, DiGeorge syndrome: the chromosome 22q11.2
deletion syndromes. Lancet. ;370(9596):1443-52.

-**Kristine Barlow (2007) :**

Traditional patterns of inheritance .autosomal recessive inheritance, Arnold text
,ED 8th ,ch.8 , pag 753-9

- **Kudva YC, Sawka AM and Young WF Jr (2003) :**

Clinical review 164: The laboratory diagnosis of adrenal pheochromocytoma: the Mayo Clinic experience. J Clin Endocrinol Metab;88(10):4533-9.

- **Kurosawa, K.; Sasaki, H.; Sato, Y.; Yamanaka, M.; Shimizu, M.; Ito, Y.;**

Okuyama, T.; Matsuo, M.; Imaizumi, K.; Kuroki, Y. and Nishimura, G

(2002) : Paternal UPD14 is responsible for a distinctive malformation complex.

Am. J. Med. Genet. 110: 268-72.

- **Iliopoulos D, Sekerli E and Vassiliou G (Jan 1 2006) :**

Patau syndrome with a long survival (146 months): a clinical report and review of literature. Am J Med Genet A. ;140(1):92-3.

-**Italiano JE and Shivdasani RA (2003) :**

Megakaryocytes and beyond: the birth of platelets . J. Thromb. Haemost. 1 (6): 1174–82.

- **Lang IA, Galloway TS and Scarlett A (September 2008) :**

Association of urinary bisphenol A concentration with medical disorders and laboratory abnormalities in adults . JAMA 300 (11): 1303–10.

- **Laurence D (2000) :**

Why are There Only Two Sexes? Proceedings: Biological Sciences, 263: 415-22.

- **Lawrence JM, Contreras R, Chen W and Sacks DA (2008) :**

Trends in the prevalence of preexisting diabetes and gestational diabetes mellitus among a racially/ethnically diverse population of pregnant women, 1999-2005. D

- **Lee H, Khan R and O'Keefe M (November 2008) :**

Aniridia: current pathology and management. Acta Ophthalmol 86 (7): 708–15.

- **Lee HJ and Chang C (2003) :**

Recent advances in androgen receptor action . Cell. Mol. Life Sci. 60 (8): 1613–22.

- **Lee YW, Lee KW and Ryu JW (2008) :**

Mutation of Glu78 of the AVP-NP_{II} gene impairs neurophysin as a carrier protein for arginine vasopressin in a family with neurohypophyseal diabetes insipidus . Ann. Clin. Lab. Sci. 38 (1): 12–4.

- **.Legro RS, Barnhart HX and Schlaff WD (2007) :**

Clomiphene, Metformin, or Both for Infertility in the Polycystic Ovary Syndrome . N Engl J Med 356 (6): 551–66

- **Lejarraga H, Caletti MG, Caino S and Jiménez A (2008) :**

Long-term growth of children with nephrogenic diabetes insipidus . Pediatr. Nephrol. 23 (11): 2007–12.

- **Lewis R (2005) :**

Human Genetics: Concepts and Applications, 6th Ed. McGraw Hill, New York.

- **Li X and Lin JF (December 2005) :**

Clinical features, hormonal profile, and metabolic abnormalities of obese women with obese polycystic ovary syndrome (in Chinese). Zhonghua Yi Xue Za Zhi 85 (46): 3266–71.

- **Lopez-Maury, L., Marguerat, S and Bahler, J (2008) :**

Tuning gene expression to changing environments: From rapid responses to evolutionary adaptation. Nature Reviews Genetics 9, 583–93.

- **Los M, Maddika S, Erb B and Schulze-Osthoff K (May 2009) :**

Switching Akt: from survival signaling to deadly response . Bioessays 31 (5): 492–5.

- **Lucchesi JC, Kelly WG and Panning B (2005) :**

Chromatin remodeling in dosage compensation . Annu. Rev. Genet. 39: 615–51

- **Lyssenko V, Jonsson A and Almgren P (November 2008) :**

Clinical risk factors, DNA variants, and the development of type 2 diabetes . N. Engl. J. Med. 359 (21): 2220–32.

Maassen JA, 't Hart LM, Janssen GM, Reiling E, Romijn JA and Lemkes HH (2006) :

Mitochondrial diabetes and its lessons for common Type 2 diabetes. Biochem. Soc. Trans. **34** (Pt 5): 819–23.

- **Maiato H, DeLuca J, Salmon E and Earnshaw W (2004) :**

The dynamic kinetochore-microtubule interface . J Cell Sci 117 (Pt 23): 5461–77.

- **Maher ER, Brueton LA and Bowdin SC (January 2003) :**

Beckwith-Wiedemann syndrome and assisted reproduction technology (ART) . Journal of medical genetics 40 (1): 62–4.

- **Maki H (2002) :**

Origins of spontaneous mutations: specificity and directionality of base-substitution, frameshift, and sequence-substitution mutageneses. Annu. Rev. Genet. 36: 279–303.

- **Marsh K and Brand-Miller J (August 2005) :**

The optimal diet for women with polycystic ovary syndrome? . Br. J. Nutr. 94 (2): 154–65.

-**Mark B (2007):**

What is a gene, post-ENCODE? History and updated definition, Genome Research 17(6): 669-81

- **Markert ML, Devlin BH and Alexieff MJ (2007) :**

Review of 54 patients with complete DiGeorge anomaly enrolled in protocols for thymus transplantation: outcome of 44 consecutive transplants. *Blood* 109 (10): 4539–47

- **Mark Stibich (August 25, 2007) :**

The Somatic Mutation Theory of Aging, About.com Health's Disease

- **Mattes, J.; Whitehead, B.; Liehr, T.; Wilkinson, I.; Bear, J.; Fagan, K.; Craven, P.; Bennetts, B. and Edwards, M (2007) :**

Paternal uniparental isodisomy for chromosome 14 with mosaicism for a supernumerary marker chromosome 14. *Am. J. Med. Genet.* 143A: 2165-71 .

- **Merke DP (Mar 2008) :**

Approach to the adult with congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *J Clin Endocrinol Metab.* ;93(3):653-60.

- **Miller, J.W.; Urbinati, C.R.; Teng-umnuay, P.; Stenberg, M.G.; Byrne, B.J.; Thornton, C.A. and Swanson (2000) :**

Recruitment of human muscleblind proteins to (CUG) n expansions associated with myotonic dystrophy. *The EMBO Journal* 19: 4439–48.

- **Miller WL, Huang N and Pandey AV (Dec 2005) :**

P450 oxidoreductase deficiency: a new disorder of steroidogenesis. *Ann N Y Acad Sci.* 1061 - 8.

- **Molitch ME. Anterior pituitary. In: Goldman L and Ausiello D (2007) :**

Cecil Medicine. 23rd ed. Philadelphia, Pa: Saunders Elsevier; chap 242.

- **Morgan RA, Dudley ME and Wunderlich JR (Oct 2006) :**

Cancer regression in patients after transfer of genetically engineered lymphocytes. *Science* 314 (5796): 126–9.

-Mortazavi A, Williams BA, McCue K, Schaeffer L and Wold B (2008) :

Mapping and quantifying mammalian transcriptomes by RNA-Seq. Nat. Methods 5: 621.

- Moser HW, Raymond GV and Dubey P (Dec 2005) :

Adrenoleukodystrophy: new approaches to a neurodegenerative disease. JAMA 294 (24): 3131–4.

- Moser, HW; Raymond GV, Lu S-E, Muenz LR, Moser AB, Xu J, Jones RO, Loes DJ, Melhem ER, Brereton NH and Odone A (2005-07) :

Follow-up of 89 asymptomatic patients with adrenoleukodystrophy treated with Lorenzo's Oil. Archives of Neurology 62 (7): p. 1073–80.

- Muller F, Sault C, Lemay C, Roussel-Mizon N, Forestier F and Frendo JL (2002) :

Second trimester two-step trisomy 18 screening using maternal serum markers. Prenat Diagn;22(7):605-8.

- Muller F, Benattar C, Roussel N, Dreux S and Cuckle H (2003) :

First-trimester screening for Down syndrome in France combining fetal nuchal translucency measurement and biochemical markers. Prenat. Diagn. 23 (10): 833–6.

- Nanninga N (2001) :

Cytokinesis in prokaryotes and eukaryotes: common principles and different solutions . Microbiol Mol Biol Rev 65 (2): 319–33.

- Nelson LM and Bakalov VK (Sep 2003) :

Mechanisms of follicular dysfunction in 46,XX spontaneous premature ovarian failure. Endocrinol Metab Clin North Am. ;32(3):613-37.

- **Neve B, Fernandez-Zapico ME and Ashkenazi-Katalan V (March 2005) :**
Role of transcription factor KLF11 and its diabetes-associated gene variants in pancreatic beta cell function . Proc. Natl. Acad. Sci. U.S.A. 102 (13): 4807–12.

- **Newell-Price J, Whiteman M, Rostami-Hodjegan A (Jan 2008) :**
Modified-release hydrocortisone for circadian therapy: a proof-of-principle study in dexamethasone-suppressed normal volunteers. Clin Endocrinol (Oxf). ;68(1):130-5.

- **Ng K, Pullirsch D, Leeb M and Wutz A (2007) :**
Xist and the order of silencing (Review Article). EMBO Rep 8: 34–9.

- **Nguyen T, Nioi P and Pickett CB (May 2009) :**
The Nrf2-antioxidant response element signaling pathway and its activation by oxidative stress. J. Biol. Chem. 284 (20): 13291–5.

- **Nicolaides KH and Wegrzyn P (2005) :**
First trimester diagnosis of chromosomal defects (in Polish). Ginekol. Pol. 76 (1): 1–8.

- **Nitsche EM and Hiort O (2000) :**
The molecular basis of androgen insensitivity. Horm. Res. 54 (5-6): 327–33.

- **Nicolaides KH, Sebire NJ, Snijders RJM and Ximenes RLS (2001) :**
Diploma in Fetal Medicine & ISUOG Educational Series: 11 - 14 weeks scan: NT and Chromosomal defects. Centrus.

- **Novosad JA, Kalantaridou SN and Nelson LM (Mar 17 2003) :**
Ovarian antibodies as detected by indirect immunofluorescence are unreliable in the diagnosis of autoimmune premature ovarian failure: a controlled evaluation. BMC Womens Health. ;3(1):2.

- **Okamoto I, Otte A, Allis C, Reinberg D and Heard E (2004) :**

Epigenetic dynamics of imprinted X inactivation during early mouse development. *Science* 303 (5658): 644–9.

-**Olson LE, Richtsmeier JT, Leszl J and Reeves RH (2004) :**

A chromosome 21 critical region does not cause specific Down syndrome phenotypes. *Science (journal)* 306 (5696): 687–90

- **Paduch DA, Fine RG, Bolyakov A and Kiper J (Nov 2008) :**

New concepts in Klinefelter syndrome. *Curr Opin Urol.* ;18(6):621-

- **Pai GS, Khan M, Barbosa E, Key LL, Craver JR, Curé JK, Betros R and Singh I (April 2000) :**

Lovastatin therapy for X-linked adrenoleukodystrophy: clinical and biochemical observations on 12 patients. *Molecular Genetics and Metabolism* 69 (4): 312–22.

- **Papageorgiou AT, Avgidou K, Spencer K, Nix B and Nicolaides KH (Feb 2006) :**

Sonographic screening for trisomy 13 at 11 to 13(+6) weeks of gestation. *Am J Obstet Gynecol.* ;194(2):397-401.

- **Patterson, D and Costa (2005) :**

Down syndrome and genetics—A case of linked histories. *Nature Reviews Genetics* 6, 137–47

- **Paul S (November 2008) :**

Dysfunction of the ubiquitin-proteasome system in multiple disease conditions: therapeutic approaches. *Bioessays* 30 (11-12): 1172–84.

-**Pearce JM (2007) :**

Pendred's syndrome. *Eur. Neurol.* 58 (3): 189–90.

- ***Pedersen SD, Brar S, Faris P and Corenblum B (2007) :***

Polycystic ovary syndrome: validated questionnaire for use in diagnosis.
Canadian family physician Médecin de famille canadien 53 (6): 1042–7.

- ***Pellegata NS, Quintanilla-Martinez L and Siggelkow H (Oct 2006) :***

Germ-line mutations in p27Kip1 cause a multiple endocrine neoplasia
syndrome in rats and humans. Proc. Natl. Acad. Sci. U.S.A. 103 (42): 15558–
63.

- ***Penman and Danny (23 August 2002) :***

Mitochondria can be inherited from both parents. NewScientist.com.

- ***Peter J. Russel (2005) :***

Genetics: A Molecular Approach. San Francisco, California, United States of
America: Pearson Education.

- ***Peters C, Charnas LR, Tan Y, Ziegler RS, Shapiro EG, DeFor T, Grewal
SS, Orchard PJ, Abel SL, Goldman AI, Ramsay NK, Dusenbery KE, Loes DJ,
Lockman LA, Kato S, Aubourg PR, Moser HW and Krivit W (2004) :*** Cerebral
X-linked adrenoleukodystrophy: the international hematopoietic cell
transplantation experience from 1982 to 1999. Blood 104 (3): 881–8.

- ***Perkins RM, Yuan CM and Welch PG (2006) :***

Dipsogenic diabetes insipidus: report of a novel treatment strategy and
literature review. Clin. Exp. Nephrol. 10 (1): 63–7.

- ***Petes, T. D (2001) :***

Meiotic recombination hot spots and cold spots. Nature Reviews Genetics 2,
360–9

- ***Petronczki, Mark; Siomos, Maria F. and Nasmyth (2003-02-21) :***

Un Ménage à Quatre The Molecular Biology of Chromosome Segregation in Meiosis, Cell 112 (4): 423-40.

- ***Pfohl-Leskowicz A and Manderville RA (2007) :***

Ochratoxin A: An overview on toxicity and carcinogenicity in animals and humans. Mol Nutr Food Res 51 (1): 61–99.

- ***Plath K, Mlynarczyk-Evans S, Nusinow D and Panning B (2002) :***

Xist RNA and the mechanism of X chromosome inactivation. Annu Rev Genet 36: 233–78.

- ***Pont SJ, Robbins JM and Bird TM (Aug 15 2006) :***

Congenital malformations among liveborn infants with trisomies 18 and 13. Am J Med Genet A. ;140(16):1749-56.

- ***Pradhan, M., Dalal, A., Khan, F. and Agrawal, S (2006) :***

Fertility in men with Down syndrome: a case report. Fertil. Steril. **86**, 1765.

- ***Purandare A, Godil MA, Prakash D (Jun 2001) :***

Spontaneous adrenal hemorrhage associated with transient antiphospholipid antibody in a child. Clin Pediatr (Phila). ;40(6):347-50.

- ***Raeder H, Johansson S and Holm PI (January 2006) :***

Mutations in the CEL VNTR cause a syndrome of diabetes and pancreatic exocrine dysfunction. Nat. Genet. **38** (1): 54–62.

- ***Raina Elley C and Kenealy T (December 2008) :***

Lifestyle interventions reduced the long-term risk of diabetes in adults with impaired glucose tolerance. Evid Based Med **13** (6): 173.

- ***Rapini, Ronald P.; Bolognia, Jean L.; Jorizzo and Joseph L (2007) :***

Dermatology: 2-Volume Set. St. Louis: Mosby. pp. 892, 4.

- *Rappold GA, Fukami M and Niesler B (2002) :*

Deletions of the homeobox gene SHOX (short stature homeobox) are an important cause of growth failure in children with short stature. J. Clin. Endocrinol. Metab. 87 (3): 1402–6.

- *Rasmussen SA, Wong LY, Yang Q and Friedman JM (2003) :*

Population-based analyses of mortality in trisomy 13 and trisomy 18. Pediatrics. ;111(4 Pt 1):777-84.

- *Raven, Peter H.; Johnson, George B.; Mason, Kenneth A.; Losos and Jonathan (2007) :*

Biology, Eighth Edition, McGraw-Hill,.

-*Rehen SK, McConnell MJ, Kaushal D, Kingsbury MA, Yang AH and Chun J (November 2001) :*

Chromosomal variation in neurons of the developing and adult mammalian nervous system. Proceedings of the National Academy of Sciences of the United States of America 98 (23): 13361–6.

-*Rehen SK, Yung YC and McCreight MP (March 2005) :*

Constitutional aneuploidy in the normal human brain. The Journal of neuroscience : the official journal of the Society for Neuroscience 25 (9): 2176–80.

- *Restivo and Angelo (February 2006) :*

22q11 Deletion syndrome: a review of some developmental biology aspects of the cardiovascular system . Journal of Cardiovascular Medicine 7 (2): 77–85.

- *R. Goswami, T. Mohapatra, N. Gupta, R. Rani, N. Tomar, A. Dikshit, and R. K. Sharma (2004) :*

Parathyroid Hormone Gene Polymorphism and Sporadic Idiopathic Hypoparathyroidism . J. Clin. Endocrinol. Metab., 89(10): 4840 - 45.

- ***Richards RI (2001) :***

Dynamic mutations: a decade of unstable expanded repeats in human genetic disease. Hum. Mol. Genet. 10 (20): 2187–94.

- ***Ricki Lewis (2003) :***

Multifactorial Traits, McGraw-Hill Higher Education,

- ***Rich SS (2006) :***

Genetics of diabetes and its complications. J. Am. Soc. Nephrol. 17 (2): 353–60.

- ***Robin NH and Shprintzen RJ (2005) :***

Defining the clinical spectrum of deletion 22q11.2. J. Pediatr. 147 (1): 90–6.

- ***Rodriguez L, Zollino M and Climent S (Jul 15 2005) :***

The new Wolf-Hirschhorn syndrome critical region (WHSCR-2): a description of a second case. Am J Med Genet A. ;136(2):175-8.

- ***Rommel N, Davidson G, Hebbard G and Omari T (2008) :***

Videomanometric evaluation of pharyngo-oesophageal dysmotility in children with velocardiofacial syndrome. J Pediatr Gastroenterol Nutr. ;46(1):87-91

- ***Rosenbusch B (November 2006) :***

The contradictory information on the distribution of non-disjunction and pre-division in female gametes. Hum. Reprod. 21 (11): 2739–42.

-***Rosyara, U. R.; Maxson-stein, K.L.; Glover, K.D.; Stein, J.M and Gonzalez-hernandez, J.L (2007) :***

Family-based mapping of FHB resistance QTLs in hexaploid wheat,
Proceedings of National Fusarium head blight forum

- **Royaux IE, Wall SM and Karniski LP (2001) :**

Pendrin, encoded by the Pendred syndrome gene, resides in the apical region of renal intercalated cells and mediates bicarbonate secretion. Proc. Natl. Acad. Sci. U.S.A. 98 (7): 4221–6.

- **Rozenberg P, Bussi res L and Chevret S (2007) :**

Screening for Down syndrome using first-trimester combined screening followed by second trimester ultrasound examination in an unselected population (in French). Gynecol Obstet Fertil 35 (4): 303–11.

- **Rubin E. and Farber J.L (2000) :**

Pathology 3rd ed., p. 225. Philadelphia: Lippincott-Raven

- **Saborio P, Hahn S, Hisano S, Latta K, Scheinman JI and Chan JC (2001) :**

Chronic renal failure: an overview from a pediatric perspective. Nephron 80 (2): 134–48 .

- **Sahoo T, del Gaudio D, German JR, Shinawi M, Peters SU, Person RE, Garnica A, Cheung SW and Beaudet AL (2008) :**

Prader-Willi phenotype caused by paternal deficiency for the HBII-85 C/D box small nucleolar RNA cluster. Nat Genet 40 (6): 719–21.

- **Saima Anwar, Saima Riazuddin, Zubair M Ahmed, Saba Tasneem1, Ateeq-ul-Jaleel, Shahid Y Khan, Andrew J Griffith, Thomas B Friedman and Sheikh Riazuddin . (2009) :**

SLC26A4 mutation spectrum associated with DFNB4 deafness and Pendred's syndrome in Pakistanis .Journal of Human Genetics advance online publication.

- ***Sakadamis, A., Angelopoulou, N., Matziari, C., Papameletiou, V. and Souftas, V (2002) :***

Bone mass, gonadal function and biochemical assessment in young men with trisomy 21. Eur. J. Obstet. Gynecol. Reprod. Biol. **100**, 208–12.

- ***Sase M, Hasegawa K and Honda R (Feb 2005) :***

Ultrasonographic findings of facial dysmorphism in Wolf-Hirschhorn syndrome. Am J Perinatol. ;22(2):99-102.

- ***Sawyer SA, Parsch J, Zhang Z and Hartl DL (2007) :***

Prevalence of positive selection among nearly neutral amino acid replacements in Drosophila . Proc. Natl. Acad. Sci. U.S.A. 104 (16): 6504–10.

- ***Schaap AH, van der Pol HG and Boer K (2002 Jul) :***

Long-term follow-up of infants after transcervical chorionic villus sampling and after amniocentesis to compare congenital abnormalities and health status. Prenat Diagn.;22(7):598-604.

- ***Scharfe C, Lu HH, Neuenburg JK, Allen EA, Li GC, Klopstock T, Cowan TM, Enns GM and Davis RW (2009) :***

Mapping gene associations in human mitochondria using clinical disease phenotypes. PLoS Comput Biol.

- ***Schwanekamp JA, Sartor MA, Karyala S, Halbleib D, Medvedovic M and Tomlinson CR (2006) :***

Genome-wide analyses show that nuclear and cytoplasmic RNA levels are differentially affected by dioxin . Biochim. Biophys. Acta 1759 (8-9): 388–402.

- ***S Das, CM Lese, M Song, JL Jensen, LA Wells, BL Barnoski, JA Roseberry, JM Camacho, DH Ledbetter and RE Schnur (2000) :***

Partial paternal uniparental disomy of chromosome 6 in an infant with neonatal diabetes, m . Am J Hum Genet 67: 1586-91.

- Seo JT, Lee JS, Oh TH and Joo KJ (Jan 2007) :

The clinical significance of bone mineral density and testosterone levels in Korean men with non-mosaic Klinefelter's syndrome. BJU Int. ;99(1):141-6.

- Shanske AL (May 1 2006) :

Trisomy 18 in a second 20-year-old woman. Am J Med Genet A. ;140(9):966-7.

- Sharquie KE, Al-Bayatti AA, Al-Ajeel AI, Al-Bahar AJ and Al-Nuaimy AA (July 2007) :

Free testosterone, luteinizing hormone/follicle stimulating hormone ratio and pelvic sonography in relation to skin manifestations in patients with polycystic ovary syndrome . Saudi Med J 28 (7): 1039–43.

- Shields, N., Taylor, N. F. and Dodd, K. J (2008) :

Effects of a community-based progressive resistance training program on muscle performance and physical function in adults with Down syndrome: a randomized controlled trial. Arch. Phys. Med. Rehabil. 89, 1215–20.

- Sieberer M, Runte I, Wilkening A, Pabst B, Ziegenbein M and Haltenhof H (May 2006) :

Spectrum of neuropsychiatric features associated with velocardiofacial syndrome (Deletion 22q11.2). Fortschr Neurol Psychiatr. ;74(5):263-74.

- Simm P and Zacharin MR (2006) :

The psychosocial impact of Klinefelter syndrome--a 10 year review. J. Pediatr. Endocrinol. Metab .19 (4): 499–505.

- ***Simpson JL and Rajkovic A (Dec 29 1999) :***

Ovarian differentiation and gonadal failure. Am J Med Genet. ;89(4):186-200.

- ***Sklar C (Jul 1999) :***

Reproductive physiology and treatment-related loss of sex hormone production. Med Pediatr Oncol. ;33(1):2-8.

- ***Skryabin BV, Gubar LV and Seeger B (2007) :***

Deletion of the MBII-85 snoRNA gene cluster in mice results in postnatal growth retardation. PLoS Genet. 3 (12):

- ***Smith CA, Katza M and Sinclair AH (2003) :***

DMRT1 Is Upregulated in the Gonads During Female-to-Male Sex Reversal in ZW Chicken Embryos. Biology of Reproduction 68: 560–70

- ***Smith JA, Vitale S and Reed GF (Feb 2004) :***

Dry eye signs and symptoms in women with premature ovarian failure. Arch Ophthalmol. ;122(2):151-6.

- ***Sniderman, AD; Bhopal R, Prabhakaran D, Sarrafzadegan N and Tchernof A (2007) :***

Why might South Asians be so susceptible to central obesity and its atherogenic consequences? The adipose tissue overflow hypothesis. International journal of epidemiology 36 (1): 220–5.

- ***Somani N, Harrison S and Bergfeld WF (2008) :***

The clinical evaluation of hirsutism. Dermatologic therapy 21 (5): 376–91.

- ***Soriano-Guillen L, Coste J, Ecosse E, Léger J, Tauber M and Cabrol S (Sep 2005) :***

Adult height and pubertal growth in Turner syndrome after treatment with recombinant growth hormone. J Clin Endocrinol Metab. ;90(9):5197-204.

- Sparber-Sauer M , M Hönig, A S Schulz, U zur Stadt, C Schütz, K M Debatin and W Friedrich (2009) :

Patients with early relapse of primary hemophagocytic syndromes or with persistent CNS involvement may benefit from immediate hematopoietic stem cell transplantation Bone Marrow , Transplantation Official journal of the EBMT Society :44, 333–8

- Speicher, M. R and Carter, N. P (2005) :

The new cytogenetics: Blurring the boundaries with molecular biology. Nature Reviews Genetics 6, 782–92

- Staple L, Andrews T, McDonald-McGinn D, Zackai E and Sullivan KE (May 2005) :

Allergies in patients with chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome) and patients with chronic granulomatous disease. Pediatr Allergy Immunol. ;16(3):226-30.

- Starke M, Albertsson Wikland K and Möller A (May-Jun 2003) :

Parents' descriptions of development and problems associated with infants with Turner syndrome: a retrospective study. J Paediatr Child Health. ;39(4):293-8.

- Stephen V. Faraone, Ming T. Tsuang and Debby W (2001) :

Genetics of mental disorders: what practitioners and students need to know. Guilford Press, Page 510

- Stratakis CA and Rennert OM (Aug 2000) :

Congenital adrenal hyperplasia: molecular genetics and alternative approaches to treatment. Crit Rev Clin Lab Sci. ;36(4):329-63.

- Stratakis CA and Rennert OM (2005) :

Turner Syndrome An Update. The Endocrinologist. ;15:27-36.

- ***Straub T and Becker PB (2007) :***

Dosage compensation: the beginning and end of generalization. *Nat Rev Genet* 8: 47–57.

-***Stuebe AM, Rich-Edwards JW, Willett WC, Manson JE and Michels KB (2005) :***

Duration of lactation and incidence of type 2 diabetes. *JAMA* 294 (20): 2601–10.

- ***Sutton, V. R.; McAlister, W. H.; Bertin, T. K.; Kaffe, S.; Wang, J.-C. C.; Yano, S.; Shaffer, L. G.; Lee, B.; Epstein, C. J and Villar, A. J (2003) :***

Skeletal defects in paternal uniparental disomy for chromosome 14 are recapitulated in the mouse model (paternal uniparental disomy 12). *Hum. Genet.* 113: 447-51 .

-***Sweeney AT, Malabanan AO and Blake MA (2000) :***

Megacolon as the presenting feature in pheochromocytoma. *J Clin Endocrinol Metab.* 85(11):3968-72.

- ***Szolar DH, KorobkinM and Reittner P (2005) :***

Adrenocortical carcinomas and adrenal pheochromocytomas: mass and enhancement loss evaluation at delayed contrast-enhanced CT. *Radiology.* 234 (2):479-85

-***Taggart R and Starr C (2006) :***

Biology The Unity and Diversity of Life: Mutated Genes and Their Protein Products. 14.4:227. Thompson Brooks/Cole

- ***Tanaka M, Nishigaki Y, Fuku N, Ibi T, Sahashi K and Koga Y (2007) :***

Therapeutic potential of pyruvate therapy for mitochondrial diseases. *Mitochondrion* 7 (6): 399–401.

- **Tarini BA (2007) :**

The current revolution in newborn screening: new technology, old controversies. Archives of pediatrics & adolescent medicine 161 (8): 767–72.

- **Terry A. Brown (2006) :**

Genomes 3. New York: Garland Science Publishing.

- **Thrasher AJ, Gaspar HB and Baum C (Sep 2006) :**

Gene therapy: X-SCID transgene leukaemogenicity". Nature 443 (7109): E5–6; discussion E6–7.

- **Tolar J, Orchard PJ, Bjoraker KJ, Ziegler RS, Shapiro EG and Charnas L (Feb 2007) :**

N-acetyl-L-cysteine improves outcome of advanced cerebral adrenoleukodystrophy. Bone Marrow Transplant 39 (4): 211–5.

- **Tong ZB and Nelson LM (Aug 1999) :**

A mouse gene encoding an oocyte antigen associated with autoimmune premature ovarian failure. Endocrinology. ;140(8):3720-6.

- **Trampal C, Engler H and Juhlin C (2004) :**

Pheochromocytomas: detection with 11C hydroxyephedrine PET. Radiology;230:423-28.

- **Tsukishiro, S.; Li, Q. Y.; Tanemura, M.; Sugiura-Ogasawara, M.; Suzumori, K. and Sonta, S (2005) :**

Paternal uniparental disomy of chromosome 14 and unique exchange of chromosome 7 in cases of spontaneous abortion. J. Hum. Genet. 50: 112-117

-Turnpenny and Peter (PDF) and Elsevier (2004). "Emery's Elements of Medical Genetics, 12th Edition, Chapter 9

- **Tüysüz, B. and Beker, D. B (2001) :**

Thyroid dysfunction in children with Down syndrome. *Acta Paediatr.* **90**, 1389–93.

- **van Kasteren YM and Schoemaker J (Sep-Oct 1999) :**

Premature ovarian failure: a systematic review on therapeutic interventions to restore ovarian function and achieve pregnancy. *Hum Reprod Update.* ;5(5):483-92.

- **van Wamel WJ, Hansenová Maňásková S, Fluit AC, Verbrugh H, de Neeling AJ, van Duijkeren E and van Belkum A (2009) :**

Short term micro-evolution and PCR-detection of methicillin-resistant and -susceptible *Staphylococcus aureus* sequence type 398. *Eur J Clin Microbiol Infect Dis*

- **Veitia RA (November 2008) :**

One thousand and one ways of making functionally similar transcriptional enhancers. *Bioessays* 30 (11-12): 1052–7.

- **Velissariou V, Christopoulou S, Karadimas C, Pihos I, Kanaka-Gantenbein C, Kapranos N, Kallipolitis G and Hatzaki A (2006) :**

Rare XXY/XX mosaicism in a phenotypic male with Klinefelter syndrome: case report. *Eur J Med Genet* 49 (4): 331- 7.

- **Virtanen S and Knip M (2003) :**

Nutritional risk predictors of beta cell autoimmunity and type 1 diabetes at a young age. *Am J Clin Nutr* 78 (6): 1053–67 .

- **von Dadelszen P, Sermer M and Hillier J (2005 May) :**

A randomised controlled trial of biopsy forceps and cannula aspiration for transcervical chorionic villus sampling. *BJOG.*;112(5):559-66.

- **Vorona E, Zitzmann M, Schuring AN and Nieschlag E (2007) :**

Clinical, endocrinological, and epigenetic features of the 46,XX male syndrome, compared with 47,XXY Klinefelter patients. J Clin Endocrinol Metab. ;92(9):3458-65.

- **Waller, D. K., Anderson, J. L., Lorey, F. and Cunningham, G. C (2000) :**

Risk factors for congenital hypothyroidism: an investigation of infant's birth weight, ethnicity, and gender in California, 1990–1998. Teratology **62**, 36–41.

- **Wangler MF, Chang AS, Feinberg AP and Debaun MR (April 2005) :**

Factors associated with preterm delivery in mothers of children with Beckwith-Wiedemann syndrome: a case cohort study from the BWS registry. American journal of medical genetics. Part A **134A** (2): 187–91.

- **Wapner RJ, Evans MI and Davis G (2002 Jun) :**

Procedural risks versus theology: chorionic villus sampling for Orthodox Jews at less than 8 weeks' gestation. Am J Obstet Gynecol.;186(6):1133- 6.

-**Watson JD, Baker TA, Bell SP, Gann A, Levine M and Losick R (2004) :**

Molecular Biology of the Gene (5th ed.). Peason Benjamin Cummings (Cold Spring Harbor Laboratory Press).

- **Wester U, Bondeson ML, Edeby C and Anneren G (2006) :**

Clinical and molecular characterization of individuals with 18p deletion: a genotype-phenotype correlation. Am J Med Genet A.;**140**:1164–71.

- **Wisniewski AB, Migeon CJ, Meyer-Bahlburg HF, Gearhart JP, Berkovitz GD, Brown TR and Money J (2000) :**

Complete androgen insensitivity syndrome: long-term medical, surgical, and psychosexual outcome". J. Clin. Endocrinol. Metab. **85** (8): 2664–9.

- Wray CJ, Rich TA, Waguespack SG, Lee JE, Perrier ND and Evans DB (2008) :

Failure to recognize multiple endocrine neoplasia 2B: more common than we think?". *Ann. Surg. Oncol.* 15 (1): 293–301

-Wysolmerski JJ and Insogna KL (2008) :

The Parathyroid Glands, Hypercalcemia, and Hypocalcemia. In: Kronenberg HM, Schlomo M, Polansky KS, Larsen PR, eds. *Williams Textbook of Endocrinology*. 11th ed. St. Louis, Mo: WB Saunders: chap. 266.

- Yamada T, Shinnoh N and Taniwaki T (September 2000) :

Lovastatin does not correct the accumulation of very long-chain fatty acids in tissues of adrenoleukodystrophy protein-deficient mice. *J. Inherit. Metab. Dis.* 23 (6): 607–14.

- Yan G, Schoenfeld D and Penney C (Apr 2000) :

Identification of premature ovarian failure patients with underlying autoimmunity. *J Womens Health Gend Based Med.* ;9(3):275-87.

- Yang AH, Kaushal D and Rehen SK (November 2003) :

Chromosome segregation defects contribute to aneuploidy in normal neural progenitor cells. *The Journal of neuroscience : the official journal of the Society for Neuroscience* 23 (32): 10454–62.

- Young WF Jr (2007) :

Clinical practice. The incidentally discovered adrenal mass. *N Engl J Med.* ;356(6):601-10.

- Zinn AR and Ross JL(Jun 2001) :

Molecular analysis of genes on Xp controlling Turner syndrome and premature ovarian failure (POF). *Semin Reprod Med.* ;19(2):141-6.