References cited in the clinical content are classified according to the type of evidence presented. The class ratings, I through V, are intended to provide a classification of the evidence but are not necessarily hierarchical. Classifications appear in parentheses at the end of each reference. References followed by an (NC) are not classified; examples include pre-published research or information from government, manufacturer, laboratory, or patient education websites.

<table>
<thead>
<tr>
<th>Classification</th>
<th>Type of Evidence</th>
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<tr>
<td>Class I</td>
<td>Meta-analysis, technology assessment, or systematic review</td>
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<tr>
<td>Class II</td>
<td>Randomized controlled trial</td>
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<td>Class III</td>
<td>Observational or epidemiologic study</td>
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<td>Class IV</td>
<td>Evidence-based guideline</td>
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<tr>
<td>Class V</td>
<td>Expert opinion, panel consensus, literature review, text or reference book, descriptive study, case report, or case series</td>
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**Class I**

Class I sources synthesize the results of multiple studies. When quantitative synthesis is possible, meta-analyses can provide a more accurate estimate of the effect or association size than individual smaller studies can. A Class I study that finds insufficient evidence to support or refute an intervention (due to a lack of appropriate primary research) is inconclusive. A potential weakness of Class I studies is that they may only assess published research, potentially leaving their findings vulnerable to publication bias.
Class II

A randomized controlled trial (RCT) is an experimental study design in which subjects are randomly assigned to an intervention or a control group. An RCT is the gold standard for testing cause and effect relationships. Intention-to-treat analysis should be performed to account for missing data points.

Class III

Observational or epidemiologic studies can suggest an association between events or findings. These associations cannot be used to establish causality. Cross-sectional, cohort, and case-control studies are all used to identify possible risk factors. Cross-sectional studies are also used to determine the prevalence of a condition. Cohort studies are used to study incidence, the natural history of a condition, prognosis after a specific exposure, and associated harms. Nonrandomized controlled trials are sometimes used when randomization is impossible or unethical.

Class IV

Evidence-based guidelines are systematically developed recommendations for clinical practice. Evidence-based guidelines identify the methodology used to gather the evidence on which the recommendations are based. Usually, a grading system for both the quality of the evidence and the strength of the recommendations is provided. Guidelines that are evidence-based may also contain consensus recommendations in areas where evidence is lacking, but these recommendations are clearly identified and appropriately graded.

Class V

Class V references may be the best information in the absence of other evidence. Expert opinion, panel consensus, literature reviews, and descriptive studies (case reports or case series) are subject to significant bias. A case series with comparison to historical controls can be plagued with missing data, and data extraction inconsistencies are common. The use of historical controls does not address how the diagnosis of disease or its treatment has evolved over time with newer technologies or medication. Textbook information may be out of date by the time the book is published.

Comparative Effectiveness Research (CER)

Citations are designated with the CER label as part of the evidence classification if the article cited is one of the following:

1. A clinical trial or other clinical study that directly compares two or more health care interventions for the same clinical scenario.
2. A systematic review that compares two or more health care interventions by synthesizing the research from previous clinical studies.
Bibliography


Ackerman et al. HRS/EHRA expert consensus statement on the state of genetic testing for the channelopathies and cardiomyopathies: this document was developed as a partnership between the Heart Rhythm Society (HRS) and the European Heart Rhythm Association (EHRA). Europace 2011. 13(8):1077-109 (V)


Ades et al. Treatment with lenalidomide does not appear to increase the risk of progression in lower risk myelodysplastic syndromes with 5q deletion. A comparative analysis by the Groupe Francophone des Myelodysplasies. Haematologica 2012. 97(2):213-8. (III)


Alavi et al. Identification of mutation in NPC2 by exome sequencing results in diagnosis of Niemann-Pick disease type C. Mol Genet Metab 2013. 110(1-2):139-44. (V)

Alba et al. HER2 status determination using RNA-ISH—a rapid and simple technique showing high correlation with FISH and IHC in 141 cases of breast cancer. Histol Histopathol 2012. 27(8):1021-7. (II)


Albright et al. Prostate cancer risk prediction based on complete prostate cancer family history. Prostate 2015. 75(4):390-8. (III)


AlesiKandarany et al. MIB1/Ki-67 labelling index can classify grade 2 breast cancer into two clinically distinct subgroups. Breast Cancer Research and Treatment 2010. (III)


American College of Medical Genetics and Genomics. Technical standards and guidelines for CFTR mutation testing Standards and guidelines for clinical genetics laboratories. Bethesda, MD: American College of Medical Genetics and Genomics; 2011. (V)


Antunes et al. Treatment algorithms in Crohn’s - up, down or something else? Best Pract Res Clin Gastroenterol 2014. 28(3):473-83. (V)


Appell et al. A skewed thiopurine metabolism is a common clinical phenomenon that can be successfully managed with a combination of low-dose azathioprine and allopurinol. J Crohns Colitis 2013. 7(6):510-3. (III)


Arese et al. Life and Death of Glucose-6-Phosphate Dehydrogenase (G6PD) Deficient Erythrocytes - Role of Redox Stress and Band 3 Modifications. Transfus Med Hemother 2012. 39(5):328-34. (V)


Arkblad et al. Multiplex ligation-dependent probe amplification improves diagnostics in spinal muscular atrophy. Neuromuscul Disord 2006. 16(12):830-8. (III)


Auer-Grumbach. Hereditary sensory neuropathy type I. Orphanet J Rare Dis 2008. 3:7. (V)


Aurilio et al. Discordant hormone receptor and human epidermal growth factor receptor 2 status in bone metastases compared to primary breast cancer. Acta Oncol 2013. 52(8):1649-56. (V)


Babushok and Bessler. Genetic predisposition syndromes: when should they be considered in the work-up of MDS? Best Pract Res Clin Haematol 2015. 28(1):55-68. (IV)


Bachanova et al. Ph+ ALL patients in first complete remission have similar survival after reduced intensity and myeloablative allogeneic transplantation: impact of tyrosine kinase inhibitor and minimal residual disease. Leukemia 2013:epub. (III)


Badens C. Variants in genetic modifiers of thalassemia can help to predict the major or intermedia type of the disease. Haematologica 2011. 96(11):1712-4. (III)


Bader et al. How and when should we monitor chimerism after allogeneic stem cell transplantation? Bone Marrow Transplant 2005. 35(2):107-19. (V)

Bader et al. Increasing mixed chimerism is an important prognostic factor for unfavorable outcome in children with acute lymphoblastic leukemia after allogeneic stem-cell transplantation: possible role for pre-emptive immunotherapy? J Clin Oncol 2004. 22(9):1696-705. (III)


Bahreini et al. A meta-analysis on concordance between immunohistochemistry (IHC) and fluorescence in situ hybridization (FISH) to detect HER2 gene overexpression in breast cancer. Breast Cancer 2014:epub. (I)


Ballinger et al. Baseline surveillance in Li-Fraumeni syndrome using whole-body magnetic resonance imaging: a meta-analysis. JAMA Oncol 2017:epub. (I)


Bar et al. Early diagnosis and care is achieved but should be improved in infants with Prader-Willi syndrome. Orphanet J Rare Dis 2017. 12(1):118. (III)


Baskin et al. Duchenne muscular dystrophy caused by a complex rearrangement between intron 43 of the DMD gene and chromosome 4. Neuromuscul Disord 2010. (III)


Batista et al. 5-FU for genital warts in non-immunocompromised individuals. Cochrane Database Syst Rev 2010(4):CD006562. (V)


Beaudenon-Huibregtse et al. Centralized molecular testing for oncogenic gene mutations complements the local cytopathologic diagnosis of thyroid nodules. Thyroid 2014. 24(10):1479-87. (III)


Berecz et al. Thioridazine steady-state plasma concentrations are influenced by tobacco smoking and CYP2D6, but not by the CYP2C9 genotype. Eur J Clin Pharmacol 2003. 59(1):45-50. (V)


Bianchi et al. The first deep intronic mutation in the NOTCH3 gene in a family with late-onset CADASIL. Neurobiol Aging 2013. 34(9):2234 e9-12. (V)


Borgiani et al. CYP4F2 genetic variant (rs2108622) significantly contributes to warfarin dosing variability in the Italian population. Pharmacogenomics 2009. 10(2):261-6. (V)


Botrel et al. Lapatinib plus chemotherapy or endocrine therapy (CET) versus CET alone in the treatment of HER-2-overexpressing locally advanced or metastatic breast cancer: systematic review and meta-analysis. Core Evid 2013. 8:69-78. (I)


Boyle et al. A CFTR corrector (lumacaftor) and a CFTR potentiator (ivacaftor) for treatment of patients with cystic fibrosis who have a phe508del CFTR mutation: a phase 2 randomised controlled trial. Lancet Respir Med 2014. 2(7):527-38. (II)


Brady et al. A detailed immunohistochemical analysis of 2 cases of papillary cystadenoma of the broad ligament: an extremely rare neoplasm characteristic of patients with von Hippel-Lindau disease. Int J Gynecol Pathol 2012. 31(2):133-40. (V)


Branstrom et al. Perceptions of genetic research and testing among members of families with an increased risk of malignant melanoma. European Journal of Cancer 2012. (III)


Bredart et al. Effect on perceived control and psychological distress of genetic knowledge in women with breast cancer receiving a BRCA1/2 test result. Breast 2017. 31:121-7. (III)


Buchanan et al. Tumor mismatch repair immunohistochemistry and DNA MLH1 methylation testing of patients with endometrial cancer diagnosed at age younger than 60 years optimizes triage for population-level germline mismatch repair gene mutation testing. J Clin Oncol 2014. 32(2):90-100. (III)


Bushby et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. Lancet Neurol 2010. 9(1):77-93. (V)


Buza et al. Toward standard HER2 testing of endometrial serous carcinoma: 4-year experience at a large academic center and recommendations for clinical practice. Mod Pathol 2013. 26(12):1605-12. (V)


Cao et al. HLA-B*58:01 allele is associated with augmented risk for both mild and severe cutaneous adverse reactions induced by allopurinol in Han Chinese. Pharmacogenomics 2012. 13(10):1193-201. (II)


Capoluongo et al. PCA3 score of 20 could improve prostate cancer detection: Results obtained on 734 Italian individuals. Clin Chim Acta 2014. 429:46-50. (III)


Carlsson and Roobol. Improving the evaluation and diagnosis of clinically significant prostate cancer in 2017. Curr Opin Urol 2017. 27(3):198-204. (V)


Carvajal et al. KIT as a therapeutic target in metastatic melanoma. JAMA 2011. 305(22):2327-34. (III)

Carvalho et al. Thiopurine-methyltransferase variants in inflammatory bowel disease: prevalence and toxicity in Brazilian patients. World J Gastroenterol 2014. 20(12):3327-34. (II)


Chakravarthy et al. Long-Term Follow-Up of a Phase II Trial of High-Dose Radiation with Concurrent 5-Fluorouracil and Cisplatin in Patients with Anal Cancer (ECOG E4292). Int J Radiat Oncol Biol Phys 2011. (II)


Chen et al. BRAF(V600E) is correlated with recurrence of papillary thyroid microcarcinoma: a systematic review, multiinstitutional primary data analysis, and meta-analysis. Thyroid 2016. 26(2):248-55. (I)


Chen et al. The association between seven ERAP1 polymorphisms and ankylosing spondylitis susceptibility: a meta-analysis involving 8,530 cases and 12,449 controls. Rheumatol Int 2012. 32(4):909-14. (I)


Chessman et al. Human leukocyte antigen class I-restricted activation of CD8+ T cells provides the immunogenetic basis of a systemic drug hypersensitivity. Immunity 2008. 28(6):822-32. (III)

Cheung et al. HLA-B alleles associated with severe cutaneous reactions to antiepileptic drugs in Han Chinese. Epilepsia 2013. (II)


Chong et al. Association of carbamazepine-induced severe cutaneous drug reactions and HLA-B*1502 allele status, and dose and treatment duration in paediatric neurology patients in Singapore. Arch Dis Child 2013;epub. (V)


Chou et al. Von Hippel-Lindau syndrome. Front Horm Res 2013. 41:30-49. (V)


Chua et al. Novel CYP2D6 and CYP2C19 variants identified in a patient with adverse reactions towards venlafaxine monotherapy and dual therapy with nortriptyline and fluoxetine. Pharmacogenet Genomics 2013. 23(9):494-7. (V)


Cincinnati Childrens Hospital Medical Center. FA Complementation and FANCD2 Western Blot. Cincinnati, OH. Available from: http://www.cincinnatichildrens.org/WorkArea/linkit.aspx?LinkIdentifier=id&ItemID=86266&libID=85954 [cited May 5 2015]. (V)


Comeglio et al. The importance of mutation detection in Marfan syndrome and Marfan-related disorders: report of 193 FBN1 mutations. Hum Mutat 2007. 28(9):928. (III)


Cook et al. Comparison of the Roche cobas® 4800 and Digene Hybrid Capture® 2 HPV tests for primary cervical cancer screening in the HPV FOCAL trial. BMC Cancer 2015. 15:968. (III CER)


Cosnes et al. Early administration of azathioprine vs conventional management of Crohn's Disease: a randomized controlled trial. Gastroenterology 2013. 145(4):758-65 e2; quiz e14-5. (II)


Couch et al. Associations between cancer predisposition testing panel genes and breast cancer. JAMA Oncol 2017:e pub. (III)


Cox et al. Evaluation of miglustat as maintenance therapy after enzyme therapy in adults with stable type 1 Gaucher disease: a prospective, open-label non-inferiority study. Orphanet J Rare Dis 2012. 7(1):102. (II)


Creutzig et al. Diagnosis and management of acute myeloid leukemia in children and adolescents: recommendations from an international expert panel. Blood 2012. 120(16):3187-205. (IV)


Crisan, ed. Hematopathology: Genomic Mechanisms of Neoplastic Diseases: Springer; 2010. (V)


Cruz-Reyes and Gamboa-Dominguez. HER2 amplification in gastric cancer is a rare event restricted to the intestinal phenotype. Int J Surg Pathol 2013. 21(3):240-6. (V)


Cui et al. BRCA2 mutations should be screened early and routinely as markers of poor prognosis: evidence from 8,986 patients with prostate cancer. Oncotarget 2017. 8(25):40222-32. (I)


Daghi and Williams. Angelman Syndrome: epub; 2015. (V)


Dalton et al. Myotonic Dystrophy Type 2. In: Pagon et al., eds. GeneReviews. Seattle (WA); 2013. (V)

D’Amico et al. Spinal muscular atrophy. Orphanet J Rare Dis 2011. 6:71. (V)


Davit-Spraul et al. Progressive familial intrahepatic cholestasis. Orphanet J Rare Dis 2009. 4:1. (V)


de Andrade et al. Higher-than-expected population prevalence of potentially pathogenic germline TP53 variants in individuals unselected for cancer history. Hum Mutat 2017:epub. (III)


De Leon. The Crucial Role of the Therapeutic Window in Understanding the Clinical Relevance of Poor Versus the Ultrarapid Metabolizer Phenotypes in Subjects Taking Drugs Metabolized by CYP2D6 or CYP2C19. J Clin Psychopharmacol 2007. 27(3):241-5. (V)

de Leon et al. DNA microarray technology in the clinical environment: the AmpliChip CYP450 test for CYP2D6 and CYP2C19 genotyping. CNS Spectr 2009. 14(1):19-34. (III)


De Luca et al. The fluctuation of PCA3 score in men undergoing first or repeat prostate biopsies. BJ U Int 2014:epub. (III)


De Napoli et al. Indeterminate single thyroid nodule: synergistic impact of mutational markers and sonographic features in triaging patients to appropriate surgery. Thyroid 2016. 26(3):390-4. (III)


Delgado et al. Chronic lymphocytic leukemia: A prognostic model comprising only two biomarkers (IGHV mutational status and FISH cytogenetics) separates patients with different outcome and simplifies the CLL-IPI. Am J Hematol 2017. 92(4):375-80. (III)


Della Porta and Malcovati. Clinical relevance of extra-hematologic comorbidity in the management of patients with myelodysplastic syndrome. Haematologica 2009. 94(5):602-6. (V)


Deng. The AlloMap genomic biomarker story: 10 years after. Clin Transplant 2017. 31(3). (V)


Di Donato et al. Update on Several/Certain Adult-Onset Genetic Leukoencephalopathies: Clinical Signs and Molecular Confirmation. J Alzheimer Dis 2014:epub. (V)


Di Leo et al. HER2 and TOP2A as predictive markers for anthracycline-containing chemotherapy regimens as adjuvant treatment of breast cancer: a meta-analysis of individual patient data. Lancet Oncol 2011. 12(12):1134-42. (I)
Di Rocco. Molecular basis and clinical management of Gaucher disease. Cardiogenetics 2013. 3(s1):e4. (V)


Dominguez-Valentin et al. Frequent mismatch-repair defects link prostate cancer to Lynch syndrome. BMC Urol 2016. 16:15. (III)

Doney et al. Lack of utility of chimerism studies obtained 2-3 months after myeloablative hematopoietic cell transplantation for ALL. Bone Marrow Transplant 2008. 42(4):271-4. (III)


Douet-Guilbert et al. Interphase FISH does not improve the detection of DEL(5q) and DEL(20q) in myelodysplastic syndromes. Anticancer Res 2011. 31(3):1007-10. (V)


Driscoll et al., Gene Reviews 2016, Prader-Willi Syndrome (V)


Drozda et al. Poor warfarin dose prediction with pharmacogenetic algorithms that exclude genotypes important for African Americans. Pharmacogenet Genomics 2015. 25(2):73-81. (III)


Dueing et al. Strategic role of frontal white matter tracts in vascular cognitive impairment: a voxel-based lesion-symptom mapping study in CADASIL. Brain 2011. 134(Pt 8):2366-75. (III)


Duh et al. A systematic review of the methods of diagnostic accuracy studies of the Afirma® Gene Expression Classifier. Thyroid 2017:epub. (I)


Durbecq et al. Topoisomerase-II alpha expression as a predictive marker in a population of advanced breast cancer patients randomly treated either with single-agent doxorubicin or single-agent docetaxel. Mol Cancer Ther 2004. 3(10):1207-14. (II)


Elia et al. The transfer of multigene panel testing for hereditary breast and ovarian cancer to healthcare: What are the implications for the management of patients and families? Oncotarget 2017. 8(2):1957-71. (III)


Eoh et al. BRCA1 and BRCA2 mutation predictions using the BRCAPRO and Myriad models in Korean ovarian cancer patients. Gynecol Oncol 2017. 145(1):137-41. (III)


Eszlinger et al. Evaluation of a two-year routine application of molecular testing of thyroid fine needle aspirations (FNA) using a 7-gene-panel in a primary referral setting in Germany. Thyroid 2017:epub. (III)

Eszlinger et al. Molecular testing of thyroid fine-needle aspirations improves presurgical diagnosis and supports the histologic identification of minimally invasive follicular thyroid carcinomas. Thyroid 2015. 25(4):401-9. (III)


European Association for the Study of the Liver. EASL Clinical Practice Guidelines: management of hepatitis C virus infection. J Hepatol 2011. 55(2):245-64 (IV)


Fang et al. Molecular characterization and copy number of SMN1, SMN2 and NAIP in Chinese patients with spinal muscular atrophy and unrelated healthy controls. BMC Musculoskeletal Disord 2015. 16:11. (II)

Fares et al. Carrier frequency of autosomal-recessive disorders in the Ashkenazi Jewish population: should the rationale for mutation choice for screening be reevaluated? Prenat Diagn 2008. 28(3):236-41. (V)


Ferder et al. Ability of VKORC1 and CYP2C9 to predict therapeutic warfarin dose during the initial weeks of therapy. J Thromb Haemost 2010. 8(1):95-100. (III)


Femandez-Mercado et al. Targeted re-sequencing analysis of 25 genes commonly mutated in myeloid disorders in del(5q) myelodysplastic syndromes. Haematologica 2013. 98(12):1856-64. (III)


Ferris et al. American Thyroid Association statement on surgical application of molecular profiling for thyroid nodules: current impact on perioperative decision making. Thyroid 2015. 25(7):760-8. (V)

Ferro et al. Prostate Health Index (Phi) and Prostate Cancer Antigen 3 (PCA3) significantly improve prostate cancer detection at initial biopsy in a total PSA range of 2-10 ng/ml. PLoS One 2013. 8(7):e67687. (III)


Fijal et al. CYP2D6 predicted metabolizer status and safety in adult patients with attention-deficit hyperactivity disorder participating in a large placebo-controlled atomoxetine maintenance of response clinical trial. J Clin Pharmacol 2015. 55(10):1167-74. (III)


Foley and Quigley. Pharmacogenomic potential of psychiatric medications and CYP2D6. MLO Med Lab Obs 2010. 42(1):32-4. (V)


Francis et al. Management guidelines for children with thyroid nodules and differentiated thyroid cancer. Thyroid 2015. 25(7):716-59. (IV)

Frank. Diagnosis and management of G6PD deficiency. Am Fam Physician 2005. 72(7):1277-82. (V)


Freeman et al. Recent advances in celiac disease. World J Gastroenterol 2011. 17(18):2259-72. (V)


Galehdari et al. Detection of a novel mutation in the GAA gene in an Iranian child with glycogen storage disease type II. Arch Iran Med 2013. 16(2):126-8. (V)


Gan et al. Natural history and management of HFE-hemochromatosis. Semin Liver Dis 2011. 31(3):293-301. (V)

Ganesan and Hussain. Question 2 Should phenytoin and carbamazepine be avoided in Asian populations with the HLA-B*1502 positive genetic variant? Arch Dis Child 2011. 96(1):104-6. (V)


Garand et al. Flow cytometry and IG/TCR quantitative PCR for minimal residual disease quantitation in acute lymphoblastic leukemia: a French multicenter prospective study on behalf of the FRALLE, EORTC and GGRAALL. Leukemia 2013. 27(2):370-6. (III)


Garrison et al. Assessing the potential cost-effectiveness of retesting IHC0, IHC1+, or FISH-negative early stage breast cancer patients for HER2 status. Cancer 2013. 119(17):3113-22. (V)


Genin et al. HLA-A*31:01 and different types of carbamazepine-induced severe cutaneous adverse reactions: an international study and meta-analysis. Pharmacogenomics J 2013:epub. (l)


Ghesquieres et al. Clinical outcome of patients with follicular lymphoma receiving chemoimmunotherapy in the PRIMA study is not affected by FCGR3A and FCGR2A polymorphisms. Blood 2012. 120(13):2650-7. (II)


Gittelman et al. PCA3 Molecular Urine Test as a Predictor of Repeat Prostate Biopsy Outcome in Men with Previous Negative Biopsies: A Prospective Multicenter Clinical Study. J Urol 2013. 190(1):64-9. (III)


Gonzales and LaSalle. The role of MeCP2 in brain development and neurodevelopmental disorders. Curr Psychiatry Rep 2010. 12(2):127-34. (V)


Gotlib and Cools. Five years since the discovery of FIP1L1-PDGFRA: what we have learned about the fusion and other molecularly defined eosinophilias. Leukemia 2008. 22(11):1999-2010. (V)


Gown et al. High concordance between immunohistochemistry and fluorescence in situ hybridization testing for HER2 status in breast cancer requires a normalized IHC scoring system. Mod Pathol 2008. 21(10):1271-7. (III)


Greenberg et al. Revised international prognostic scoring system for myelodysplastic syndromes. Blood 2012. 120(12):2454-65. (V)


Grindedal et al. Current guidelines for BRCA testing of breast cancer patients are insufficient to detect all mutation carriers. BMC Cancer 2017. 17(1):438. (III)

Grisedi et al. A common haplotype at the 5' end of the RET proto-oncogene, overrepresented in Hirschsprung patients, is associated with reduced gene expression. Hum Mutat 2005. 25(2):189-95. (III)


Guo et al. Lapatinib, a dual inhibitor of epidermal growth factor receptor and human epidermal growth factor receptor 2, potentiates the antitumor effects of cisplatin on esophageal carcinoma. Dis Esophagus 2013. 26(5):487-95. (V)

Guo et al. Wide mutation spectrum and frequent variant Ala27Thr of FBN1 identified in a large cohort of Chinese patients with sporadic TAAD. Sci Rep 2015. 5:13115. (III)


Haberle et al. Suggested Guidelines for the Diagnosis and Management of Urea Cycle Disorders. Orphanet J Rare Dis 2012. 7(1):32. (V)


Hall et al. Familial adenomatous polyposis. Orphanet J Rare Dis 2009. 4:22. 2009. (V)


Hall-Flavin et al. Using a pharmacogenomic algorithm to guide the treatment of depression. Transl Psychiatry 2012. 2:e172. (III)


Harteveld and Higgs. Alpha-thalassaemia. Orphanet J Rare Dis 2010. 5:13 (V)


Haugen et al. 2015 American Thyroid Association management guidelines for adult patients with thyroid nodules and differentiated thyroid cancer: the American Thyroid Association Guidelines Task Force on Thyroid Nodules and Differentiated Thyroid Cancer. Thyroid 2016. 26(1):1-133. (IV)


Hehlmann et al. Deep molecular response is reached by the majority of patients treated with imatinib, predicts survival, and is achieved more quickly by optimized high-dose imatinib: results from the randomized CML-study IV. J Clin Oncol 2014. 32(5):415-23. (II)


Hershberger et al. Progress with genetic cardiomyopathies: screening, counseling, and testing in dilated, hypertrophic, and arrhythmogenic right ventricular dysplasia cardiomyopathy. Circ Heart Fail 2009. 2(3):253-61. (V)


Hicks and Whitney-Miller. The evolving role of HER2 evaluation for diagnosis and clinical decision making for breast and gastric adenocarcinoma. Biotech Histochem 2013. 88(3-4):121-31. (V)


Hillman et al. How does altering the resolution of chromosomal microarray analysis in the prenatal setting affect the rates of pathological and uncertain findings? J Matern Fetal Neonatal Med 2014. 27(7):649-57. (V)


Hoffmann et al. Treatment and outcome of 2904 CML patients from the EUTOS population-based registry. Leukemia 2016:epub. (III)


Hollink et al. Favorable prognostic impact of NPM1 gene mutations in childhood acute myeloid leukemia, with emphasis on cytogenetically normal AML. Leukemia 2009. 23(2):262-70. (III)


Hospira. Methotrexate Injection, USP 2011 (NC)


Hu et al. HER2 amplification, overexpression and score criteria in esophageal adenocarcinoma. Mod Pathol 2011. 24(7):899-907. (III)


Hulot et al. Cardiovascular risk in clopidogrel-treated patients according to cytochrome P450 2C19*2 loss-of-function allele or proton pump inhibitor coadministration: a systematic meta-analysis. J Am Coll Cardiol 2010. 56(2):134-43. (I)


Huynh et al. The clinical and biochemical spectrum of congenital adrenal hyperplasia secondary to 21-hydroxylase deficiency. The Clinical Biochemist Reviews / Australian Association of Clinical Biochemists 2009. 30(2):75-86 (V)


Ibrahim et al. Methylation analysis and diagnostics of Beckwith-Wiedemann syndrome in 1,000 subjects. Clin Epigenetics 2014. 6(1):11. (III)


Iftner et al. Head-to-head comparison of the RNA-based Aptima human papillomavirus (HPV) assay and the DNA-based Hybrid Capture 2 HPV test in a routine screening population of women aged 30 to 60 years in Germany. J Clin Microbiol 2015. 53(8):2509-16. (III CER)


Iourov et al. Xq28 (MECP2) microdeletions are common in mutation-negative females with Rett syndrome and cause mild subtypes of the disease. Mol Cytogenet 2013. 6(1):53. (V)


Itzhari et al. Chromosomal minimal critical regions in therapy-related leukemia appear different from those of de novo leukemia by high-resolution aCGH. PLoS One 2011. 6(2):e16623. (V)


Jabeen et al. Impact of genetic variants of RFC1, DHFR and MTHFR in osteosarcoma patients treated with high-dose methotrexate. Pharmacogenomics J 2015 (II)


James et al. Large genomic rearrangements in the familial breast and ovarian cancer gene BRCA1 are associated with an increased frequency of high risk features. Fam Cancer 2015. 14(2):287-95. (III)


Jang et al. Usefulness of NRAS codon 61 mutation analysis and core needle biopsy for the diagnosis of thyroid nodules previously diagnosed as atypia of undetermined significance. Endocrine 2016. 52(2):305-12. (III)


Jara et al. Utility of BRAF mutation detection in fine-needle aspiration biopsy samples read as "suspicious for papillary thyroid carcinoma". Head Neck 2015. 37(12):1788-93. (III)

Jasperson and Burt. APC-Associated Polyposis Conditions 2014. (V)


Khera and Ghuliani. Type 0 spinal muscular atrophy with multisystem involvement. Indian Pediatr 2014. 51(11):923-4. (V)


Kim et al. ABCB1, FGCR2A, and FGCR3A polymorphisms in patients with HER2-positive metastatic breast cancer who were treated with first-line taxane plus trastuzumab chemotherapy. Oncology 2012. 83(4):218-27. (II)


Kirchheiner et al. Pharmacogenetics of antidepressants and antipsychotics: the contribution of allelic variations to the phenotype of drug response. Mol Psychiatry 2004. 9(5):442-73. (I)


Klein et al. A genomic classifier improves prediction of metastatic disease within 5 years after surgery in node-negative high-risk prostate cancer patients managed by radical prostatectomy without adjuvant therapy. Eur Urol 2015. 67(4):778-86. (III)


Kloos et al. A genomic alternative to identify medullary thyroid cancer preoperatively in thyroid nodules with indeterminate cytology. Thyroid 2016. 26(6):785-93. (III)


Knoop et al. retrospective analysis of topoisomerase IIa amplifications and deletions as predictive markers in primary breast cancer patients randomly assigned to cyclophosphamide, methotrexate, and fluorouracil or cyclophosphamide, epirubicin, and fluorouracil: Danish Breast Cancer Cooperative Group. J Clin Oncol 2005. 23(30):7483-90. (III)


Kohmann and Gruber. Lynch syndrome: Gene Reviews; 2014. (V)


Kowalska et al. The usefulness of determining the presence of BRAF V600E mutation in fine-needle aspiration cytology in indeterminate cytological results. Endokrynol Pol 2016. 67(1):41-7. (III)


Kransdorf EP. Genetic and genomic approaches to the detection of heart transplant rejection. Personalized Medicine 2012. 9(7):693-705. (V)

Kraus et al. Gene panel sequencing in familial breast/ovarian cancer patients identifies multiple novel mutations also in genes others than BRCA1/2. Int J Cancer 2017. 140(1):95-102. (III)


Kroger et al. NCI First International Workshop on the Biology, Prevention, and Treatment of Relapse after Allogeneic Hematopoietic Stem Cell Transplantation: report from the Committee on Disease-Specific Methods and Strategies for Monitoring Relapse following Allogeneic Stem Cell Transplantation. Part I: Methods, acute leukemias, and myelodysplastic syndromes. Biol Blood Marrow Transplant 2010. 16(9):1187-211. (V)


Kvendgen et al. Lenalidomide does not increase AML progression risk in RBC transfusion-dependent patients with Low- or Intermediate-1-risk MDS with del(5q): a comparative analysis. Leukemia 2013. 27(5):1072-9. (III)


Kumarasinghe et al. HER2 status in gastric/gastro-oesophageal junctional cancers: should determination of gene amplification by SISH use HER2 copy number or HER2: CEP17 ratio? Pathology 2014. 46(3):184-7. (V)


Kwon et al. Early peripheral blood and T-cell chimerism dynamics after umbilical cord blood transplantation supported with haploidentical cells. Bone Marrow Transplant 2014. 49(2):212-8. (V)


Lacchetti et al. Routine HPV testing in head and neck squamous cell carcinoma. Toronto: Cancer Care Ontario; 2013. (IV)


Lassuthova et al. Improving diagnosis of inherited peripheral neuropathies through gene panel analysis. Orphanet J Rare Dis 2016. 11(1):118. (III)

Lastra et al. Implications of a suspicious Afirma test result in thyroid fine-needle aspiration cytology: an institutional experience. Cancer Cytopathol 2014. 122(10):737-44. (III)


Lawler et al. Serial chimerism analyses indicate that mixed haemopoietic chimerism influences the probability of graft rejection and disease recurrence following allogeneic stem cell transplantation (SCT) for severe aplastic anaemia (SAA): indication for routine assessment of chimerism post SCT for SAA. Br J Haematol 2009. 144(6):933-45. (III)


Lech et al. Colorectal cancer tumour markers and biomarkers: Recent therapeutic advances. World J Gastroenterol 2016. 22(5):1745-55. (V)


Lee et al. CAG repeat expansion in Huntington disease determines age at onset in a fully dominant fashion. Neurology 2012. 78(10):690-5. (V)


Lee et al. Evaluation of a genomic classifier in radical prostatectomy patients with lymph node metastasis. Res Rep Urol 2016. 8:77-84. (III)


Li et al. Aspirin use after diagnosis but not prediagnosis improves established colorectal cancer survival: a meta-analysis. Gut 2015. 64(9):1419-25. (I)

Li et al. BRAFV600E mutation in papillary thyroid microcarcinoma: a meta-analysis. Endocr Relat Cancer 2015. 22(2):159-68. (I)
Li et al. Effectiveness of prophylactic surgeries in BRCA1 or BRCA2 mutation carriers: a meta-analysis and systematic review. Clin Cancer Res 2016:epub. (I)


Li et al. Spectrum of UGT1A1 Variations in Chinese Patients with Crigler-Najjar Syndrome Type II. PLoS One 2015. 10(5):e0126263. (V)


Lin et al. Validation and workflow optimization of human epidermal growth factor receptor 2 testing using INFORM HER2 dual-color in situ hybridization. Hum Pathol 2013. 44(11):2590-6. (V)


Lin et al. Screening for Cognitive Impairment in Older Adults: An Evidence Update for the U.S. Preventive Services Task Force. In: Screening for Cognitive Impairment in Older Adults: An Evidence Update for the U.S. Preventive Services Task Force. Rockville (MD); 2013. (I)


Lindstrom et al. Clinically used breast cancer markers such as estrogen receptor, progesterone receptor, and human epidermal growth factor receptor 2 are unstable throughout tumor progression. J Clin Oncol 2012. 30(21):2601-8. (II)


Lion et al. The EuroChimerism concept for a standardized approach to chimerism analysis after allogeneic stem cell transplantation. Leukemia 2012. 26(8):1821-8. (V)


Lipsyc and Yaeger. Impact of somatic mutations on patterns of metastasis in colorectal cancer. J Gastrointest Oncol 2015. 6(6):645-9. (V)

List et al. Extended survival and reduced risk of AML progression in erythroid-responsive lenalidomide-treated patients with lower-risk del(5q) MDS. Leukemia 2013:epub. (II)


Liu et al. Examination of multiple UGT1A and DPYD polymorphisms has limited ability to predict the toxicity and efficacy of metastatic colorectal cancer treated with irinotecan-based chemotherapy: a retrospective analysis. BMC Cancer 2017. 17(1):437. (III)


Liu et al. What we have learned from the next-generation sequencing: Contributions to the genetic diagnosis and understanding of pathomechanisms of neurodegenerative diseases. J Neurogenet 2015. 29(2-3):103-12. (V)


Llerena et al. QTc interval, CYP2D6 and CYP2C9 genotypes and risperidone plasma concentrations. J Psychopharmacol 2004. 18(2):189-93. (V)


Locharemkul et al. Carbamazepine and phenytoin induced Stevens-Johnson syndrome is associated with HLA-B*1502 allele in Thai population. Epilepsia 2008. 49(12):2087-91. (III)


Loovers and van der Weide. Implementation of CYP2D6 genotyping in psychiatry. Expert Opin Drug Metab Toxicol 2009. 5(9):1065-77. (III)


Lorson and Lorson. SMN-inducing compounds for the treatment of spinal muscular atrophy. Future Med Chem 2012. 4(16):2067-84. (V)


Ma et al. Advances in pharmacogenomics of antiretrovirals: an update. Pharmacogenomics 2007. 8(9):1169-78. (V)


Mai et al. Prevalence of cancer at baseline screening in the National Cancer Institute Li-Fraumeni syndrome cohort. JAMA Oncol 2017:epub. (III)


Maron. A paradigm shift in our understanding of the development of the hypertrophic cardiomyopathy phenotype?: not so fast! Circulation 2013. 127(1):10-2. (V)


Marquard and Eng. Multiple Endocrine Neoplasia Type 2, In: GeneReviews. 2015: epub (V)


Martignano et al. GSTP1 Methylation and Protein Expression in Prostate Cancer: Diagnostic Implications. Dis Markers 2016. 2016:4358292. (III)


Mehanna et al. Analysis of CYP2D6 genotype and response to tetrabenazine. Mov Disord 2013. 28(2):210-5. (III)

Mehrotra et al. Quantitative, spatial resolution of the epigenetic field effect in prostate cancer. Prostate 2008. 68(2):152-60. (III)


Mersch et al. Cancers associated with BRCA1 and BRCA2 mutations other than breast and ovarian. Cancer 2015. 121(2):269-75. (III)


Mickelson et al. Comparison of donor chimerism following myeloablative and nonmyeloablative alogeneic hematopoietic SCT. Bone Marrow Transplant 2011. 46(1):84-9. (III)


Min toda-Vilela. Role of polymorphisms in factor V (FV Leiden), prothrombin, plasminogen activator inhibitor type-1 (PAI-1), methylenetetrahydrofolate reductase (MTHFR) and cystathionine beta-synthase (CBS) genes as risk factors for thrombophilias. Mini Rev Med Chem 2012. 12(10):997-1006. (V)


Mitrovic et al. Correlation between ER, PR, HER-2, Bcl-2, p53, proliferative and apoptotic indexes with HER-2 gene amplification and TOP2A gene amplification and deletion in four molecular subtypes of breast cancer. Target Oncol 2013:epub. (III)


Moller et al. Heterogeneous patterns of DNA methylation-based field effects in histologically normal prostate tissue from cancer patients. Sci Rep 2017. 7:40636. (III)


Mooorthie et al. Systematic review and meta-analysis to estimate the birth prevalence of five inherited metabolic diseases. J Inherit Metab Dis 2014;epub. (I)


Morel et al. Clinical relevance of different dihydropyrimidine dehydrogenase gene single nucleotide polymorphisms on 5-fluorouracil tolerance. Mol Cancer Ther 2006. 5:2895-904. (III)

Moreton et al. Changing clinical patterns and increasing prevalence in CADASIL. Acta Neurol Scand 2014;epub. (II)

Moretti et al. TOP2A protein by quantitative immunofluorescence as a predictor of response to epirubicin in the neoadjuvant treatment of breast cancer. Future Oncol 2013. 9(10):1477-87. (III)


Munch-Petersen et al. Reliability of histological malignancy grade, ER and HER2 status on core needle biopsy vs surgical specimen in breast cancer. APMIS 2014. 122(9):750-4. (V)


Munoz-Couselo et al. Recent advances in the treatment of melanoma with BRAF and MEK inhibitors. Ann Transl Med 2015. 3(15):207. (V)


Myers et al. Proteomic Upregulation of Fatty Acid Synthase and Fatty Acid Binding Protein 5 and Identification of Cancer- and Race-Specific Pathway Associations in Human Prostate Cancer Tissues. J Cancer 2016. 7(11):1452-64. (III)


Mysliwiec et al. Pheochromocytoma--analysis of 15 consecutive cases from one centre. Endokrynol Pol 2013. 64(3):192-6. (III)


Nahas et al. Molecular Testing in Myelodysplastic Syndromes for the Practicing Oncologist: Will the Progress Fulfill the Promise? Oncologist 2015. 20(9):1069-76. (V)


Nevoral et al. Symptom positivity is essential for omitting biopsy in children with suspected celiac disease according to the new ESPGHAN guidelines. Eur J Pediatr 2013:epub. (V)

Newton et al. Tumour MLH1 promoter region methylation testing is an effective prescreen for Lynch Syndrome (HNPCC). J Med Genet 2014. 51(12):789-96. (III)


Ngeow et al. Prevalence of germline PTEN, BMPR1A, SMAD4, STK11, and ENG mutations in patients with moderate-load colorectal polyps. Gastroenterology 2013. 144(7):1402-9, 9 e1-5. (III)


Nielsen et al. Lack of independent prognostic and predictive value of centromere 17 copy number changes in breast cancer patients with known HER2 and TOP2A status. Mol Oncol 2012. 6(1):88-97. (III)


Nikiforov et al. Highly accurate diagnosis of cancer in thyroid nodules with follicular neoplasm/suspicious for a follicular neoplasm cytology by ThyroSeq v2 next-generation sequencing assay. Cancer 2014. 120(23):3627-34. (III)


Nogi et al. [Which biomarkers can we use?]. Nihon Rinsho 2012. 70(5):816-20. (V)


Nordestgaard et al. Familial hypercholesterolaemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease: Consensus Statement of the European Atherosclerosis Society. Eur Heart J 2013:eupub. (V)


Nusliha et al. Congenital hypertrophy of retinal pigment epithelium (CHRPE) in patients with familial adenomatous polyposis (FAP); a polyposis registry experience. BMC Res Notes 2014. 7:734. (III)


Ogilvie et al. The future of prenatal diagnosis: rapid testing or full karyotype? An audit of chromosome abnormalities and pregnancy outcomes for women referred for Down's Syndrome testing. BJOG 2005. 112(10):1369-75. (III)


Oh and Hustead. Causes and evaluation of mildly elevated liver transaminase levels. Am Fam Physician 2011. 84(9):1003-8. (V)

Ohnishi et al. Chronic eosinophilic leukaemia with FIP1L1-PDGFRα fusion and T6741 mutation that evolved from Langerhans cell histiocytosis with eosinophilia after chemotherapy. Br J Haematol 2006. 134(5):547-9. (V)


Ok et al. TP53 mutation characteristics in therapy-related myelodysplastic syndromes and acute myeloid leukemia is similar to de novo diseases. J Hematol Oncol 2015. 8:45. (V)


O’Neill et al. Primary care providers’ willingness to recommend BRCA1/2 testing to adolescents. Fam Cancer 2010. 9(1):43-50. (V)


Orvieto. Preimplantation genetic screening- the required RCT that has not yet been carried out. Reprod Biol Endocrinol 2016. 14(1):35. (V)

O’Shea et al. Next generation sequencing is informing phenotype: a TP53 example. Fam Cancer 2017:epub. (III)


Overgaard et al. TP53 mutation is an independent prognostic marker for poor outcome in both node-negative and node-positive breast cancer. Acta Oncol 2000. 39(3):327-33. (III)


Pajor et al. Urovysion: Considerations on modifying current evaluation scheme, including immunophenotypic targeting and locally set, statistically derived diagnostic criteria. Cytometry A 79(5):375-82. (III)


Pappachan et al. Diagnosis and management of pheochromocytoma: a practical guide to clinicians. Curr Hypertens Rep 2014. 16(7):442. (V)


Pardanani. How I treat patients with indolent and smoldering mastocytosis (rare conditions but difficult to manage). Blood 2013. 121(16):3085-94. (V)


Parikh et al. Should IGHV status and FISH testing be performed in all CLL patients at diagnosis? A systematic review and meta-analysis. Blood 2016. 127(14):1752-60. (I)


Patterson et al. Disease and patient characteristics in NP-C patients: findings from an international disease registry. Orphanet J Rare Dis 2013. 8:12. (V)


Penas-Lledo et al. CYP2D6 ultrarapid metabolism and early dropout from fluoxetine or amitriptyline monotherapy treatment in major depressive patients. Mol Psychiatry 2013. 18(1):8-9. (V)


Persky et al. Fc gamma receptor 3a genotype predicts overall survival in follicular lymphoma patients treated on SWOG trials with combined monoclonal antibody plus chemotherapy but not chemotherapy alone. Haematologica 2012. (III)


Pescia et al. Cell-free DNA testing of an extended range of chromosomal anomalies: clinical experience with 6,388 consecutive cases. Genet Med 2016. (V)


Pillers. A new day for Duchenne's?: The time has come for newborn screening. Mol Genet Metab 2014;epub. (V)


Pirmohamed. Genetic factors in the predisposition to drug-induced hypersensitivity reactions. AAPSJ 2006. 8(1):E20-6. (V)


Pitchford et al. Fluorescence in situ hybridization testing for -5/5q, -7/7q, 8, and del(20q) in primary myelodysplastic syndrome correlates with conventional cytogenetics in the setting of an adequate study. Am J Clin Pathol 2010. 133(2):260-4. (III)


Polley et al. An international study to increase concordance in Ki67 scoring. Mod Pathol 2015. (V)


Port et al. Prognostic significance of FLT3 internal tandem duplication, nucleophosmin 1, and CEBPA gene mutations for acute myeloid leukemia patients with normal karyotype and younger than 60 years: a systematic review and meta-analysis. Ann Hematol 2014. 93(8):1279-86. (I)

Pospisilova et al. ERIC recommendations on TP53 mutation analysis in chronic lymphocytic leukemia. Leukemia 2012. 26(7):1458-61. (V)


Pringsheim et al. The incidence and prevalence of Huntington's disease: a systematic review and meta-analysis. Mov Disord 2012. 27(9):1083-91. (I)


Radkay et al. Thyroid nodules with KRAS mutations are different from nodules with NRAS and HRAS mutations with regard to cytopathologic and histopathologic outcome characteristics. Cancer Cytopathol 2014. 122(12):873-82. (III)


Ramsden et al. Practice guidelines for the molecular analysis of Prader-Willi and Angelman syndromes. BMC Med Genet 2010. 11:70. (V)


Renwick et al. ATM mutations that cause ataxia-telangiectasia are breast cancer susceptibility alleles. Nat Genet 2006. 38(8):873-5. (IV)


Reyes et al. Presymptomatic genetic testing in CADASIL. J Neurol 2012. 259(10):2131-6 (III)


Rodriguez-Reyenga L. Motor and mental dysfunction in mother-daughter transmitted FXTAS. Neurology 2010. 75(15):1370-6. (V)


Rogers et al. CYP2D6 genotype information to guide pimozide treatment in adult and pediatric patients: basis for the U.S. Food and Drug Administration's new dosing recommendations. J Clin Psychiatry 2012. 73(9):1187-90. (V)


Romanos et al. Improving coeliac disease risk prediction by testing non-HLA variants additional to HLA variants. Gut 2013. 63(3):415-22. (V)


Rosenow et al. CD34(+) lineage specific donor cell chimerism for the diagnosis and treatment of impending relapse of AML or myelodysplastic syndrome after allo-SCT. Bone Marrow Transplant 2013. 48(8):1070-6. (V)


Rossmann et al. A candidate gene study of capecitabine-related toxicity in colorectal cancer identifies new toxicity variants at DPYD and a putative role for ENOSF1 rather than TYMS. Gut 2014:epub. (II)


Rossi et al. Relevance of BRAF(V600E) mutation testing versus RAS point mutations and RET/PTC rearrangements evaluation in the diagnosis of thyroid cancer. Thyroid 2015. 25(2):221-8. (III)


Sampaolo et al. Distinct disease phenotypes linked to different combinations of GAA mutations in a large late-onset GSDII sibship. Orphanet J Rare Dis 2013. 8(1):159. (V)


Sanders et al. Structural Fetal Abnormalities: The Total Picture, 2 edn. St. Louis: Mosby; 2002. (V)


Saya et al. Baseline results from the UK SIGNIFY study: a whole-body MRI screening study in TP53 mutation carriers and matched controls. Fam Cancer 2017. 16(3):433-40. (III)


Scattoni et al. Head-to-head comparison of prostate health index and urinary PCA3 for predicting cancer at initial or repeat biopsy. J Urol 2013. 190(2):496-501. (III)


Schnittger et al. KIT-D816 mutations in AML1-ETO-positive AML are associated with impaired event-free and overall survival. Blood 2006. 107(5):1791-9. (III)

Schnyder et al. HLA-B*57:01(+)-abacavir-naive individuals have specific T cells but no patch test reactivity. J Allergy Clin Immunol 2013. 132(3):756-8. (V)


Schroder et al. Screening for prostate cancer decreases the risk of developing metastatic disease: findings from the European Randomized Study of Screening for Prostate Cancer (ERSPC). Eur Urol 2012. 62(5):745-52. (III)


Scordo et al. Cytochrome P450 2D6 genotype and steady state plasma levels of risperidone and 9-hydroxyrisperidone. Psychopharmacology (Berl) 1999. 147(3):300-5. (V)


Seo et al. Additional BRAF mutation analysis may have additional diagnostic value in thyroid nodules with "suspicious for malignant" cytology alone even when the nodules do not show suspicious US features. Endocrine 2014:epub. (III)


Shiba et al. DNMT3A mutations are rare in childhood acute myeloid leukaemia, myelodysplastic syndromes and juvenile myelomonocytic leukaemia. Br J Haematol 2012. 156(3):413-4. (III)
Shields et al. High resolution mapping of the binding site on human IgG1 for Fc gamma RII, Fc gamma RIII, and FcRn and design of IgG1 variants with improved binding to the Fc gamma R. The Journal of Biological Chemistry 2001. 276(9):6591-604. (V)


Shiller. Laboratory Methods for the Diagnosis of Hereditary Amyloidoses: InTech; 2011. (V)


Shrestha et al. Correlation between histological diagnosis and mutational panel testing of thyroid nodules: a two-year institutional experience. Thyroid 2016. 26(8):1068-76. (III)


Shuldiner et al. Association of cytochrome P450 2C19 genotype with the antiplatelet effect and clinical efficacy of clopidogrel therapy. JAMA 2009. 302(8):849-57. (III)


Smith and Hung. The dilemma of diagnostic testing for Prader-Willi syndrome. Transl Pediatr 2017. 6(1):46-56. (V)


Sorich et al. CYP2C19 genotype has a greater effect on adverse cardiovascular outcomes following percutaneous coronary intervention and in Asian populations treated with clopidogrel: a meta-analysis. Circ Cardiovasc Genet 2014. 7(6):895-902. (I)


Southey et al. PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. J Med Genet 2016: epub. (III)

Soverini et al. Drug resistance and BCR-ABL kinase domain mutations in Philadelphia chromosome-positive acute lymphoblastic leukemia from the imatinib to the second-generation tyrosine kinase inhibitor era: The main changes are in the type of mutations, but not in the frequency of mutation involvement. Cancer 2014. 120(7):1002-9. (III)


Spinney. Uncovering the true prevalence of Huntington’s disease. Lancet Neurol 2010. 9(8):760-1. (V)


Stamp et al. The use of low dose methotrexate in rheumatoid arthritis - are we entering a new era of therapeutic drug monitoring and pharmacogenomics? Biomed Pharmacother 2006. 60(10):678-87. (V)

Stampfer et al. Niemann-Pick disease type C clinical database: cognitive and coordination deficits are early disease indicators. Orphanet J Rare Dis 2013. 8:35. (V)


Stott-Miller et al. HOXB13 mutations in a population-based, case-control study of prostate cancer. Prostate 2013. 73(6):634-41. (III)


Strehle and Straub. Recent advances in the management of Duchenne muscular dystrophy. Arch Dis Child 2015. 100(12):1173-7. (V)


Su et al. Association of telomerase reverse transcriptase promoter mutations with clinicopathological features and prognosis of thyroid cancer: a meta-analysis. Onco Targets Ther 2016. 9:6965-76. (I)


Su et al. Radiation exposure, young age, and female gender are associated with high prevalence of RET/PTC1 and RET/PTC3 in papillary thyroid cancer: a meta-analysis. Oncotarget 2016. 7(13):16716-30. (I)


Susswein et al. Pathogenic and likely pathogenic variant prevalence among the first 10,000 patients referred for next-generation cancer panel testing. Genet Med 2015. 18(8):823-32. (III)


Tangamornsuk et al. Relationship between the HLA-B*1502 allele and carbamazepine-induced Stevens-Johnson syndrome and toxic epidermal necrolysis: a systematic review and meta-analysis. JAMA Dermatol 2013. 149(9):1025-32. (I)


Tassone et al. FMR1 CGG allele size and prevalence ascertained through newborn screening in the United States. Genome Med 2012. 4(12):100. (III)


Te Loo et al. Is there a role for the MTHFR 677C>T and 1298A>C polymorphisms in methotrexate-induced liver toxicity? Pharmacogenomics 2014. 15(11):1401-3 (V)


Tedaldi et al. Multiple-gene panel analysis in a case series of 255 women with hereditary breast and ovarian cancer. Oncotarget 2017:epub. (III)


Therkildsen et al. The predictive value of KRAS, NRAS, BRAF, PIK3CA and PTEN for anti-EGFR treatment in metastatic colorectal cancer: A systematic review and meta-analysis. Acta Oncol 2014. 53(7):852-64 (I)


Thomas. Functional muscle ischemia in Duchenne and Becker muscular dystrophy. Front Physiol 2013. 4:381. (V)


Thomas et al. Mipomersen, an Apolipoprotein B Synthesis Inhibitor, Reduces Atherogenic Lipoproteins in Patients with Severe Hypercholesterolemia at High Cardiovascular Risk: A Randomized, Double-Blind, Placebo-Controlled Trial. J Am Coll Cardiol 2013:epub. (II)


Tikka. Diagnosing Vascular Dementia by Skin Biopsy - Uniqueness of CADASIL. In: Khopkar, ed. Skin Biopsy-Perspectives: InTech; 2011. (V)


Treon et al. Attainment of complete/very good partial response following rituximab-based therapy is an important determinant to progression-free survival, and is impacted by polymorphisms in FCGR3A in Waldenstrom macroglobulinaemia. British Journal of Haematology 2011. 154(2):223-8. (III)


Trimboli et al. Use of fine-needle aspirate calcitonin to detect medullary thyroid carcinoma: A systematic review. Diagn Cytopathol 2016. 44(1):45-51. (I)


Trock et al. Evaluation of GSTP1 and APC methylation as indicators for repeat biopsy in a high-risk cohort of men with negative initial prostate biopsies. BJU Int 2012. 110(1):56-62. (III)


Turatti et al. Short communication: UGT1A1*28 variant allele is a predictor of severe hyperbilirubinemia in HIV-infected patients on HAART in southern Brazil. AIDS Res Hum Retroviruses 2012. 28(9):1015-8. (V)


Twigg et al. Cellular interference in craniofrontonasal syndrome: males mosaic for mutations in the X-linked EFNB1 gene are more severely affected than true hemizygotes. Hum Mol Genet 2013. 22(8):1654-62. (V)


Tziotou et al. Polymorphisms of uridine glucuronosyltransferase gene and irinotecan toxicity: low dose does not protect from toxicity. Ecancermedicalscience 2014. 8:428. (II)


InterQual® Molecular Diagnostics Criteria


Use of preimplantation genetic diagnosis for serious adult onset conditions: a committee opinion. Fertil Steril 2013. 100(1):54-7. (V)


Vaananen et al. Association of transcript levels of 10 established or candidate-biomarker gene targets with cancerous versus non-cancerous prostate tissue from radical prostatectomy specimens. Clin Biochem 2013. 46(7-8):670-4. (III)


Van Cutsem et al. HER2 screening data from ToGA: targeting HER2 in gastric and gastroesophageal junction cancer. Gastric Cancer 2014:epub. (V)


van den Hout et al. The natural course of infantile Pompe's disease: 20 original cases compared with 133 cases from the literature. Pediatrics 2003. 112(2):332-40. (III)


Van Neste et al. Risk score predicts high-grade prostate cancer in DNA-methylation positive, histopathologically negative biopsies. Prostate 2016. 76(12):1078-87. (III)


Vandenberghhe et al. Clinical and molecular features of FIP1L1-PDGFRA (+) chronic eosinophilic leukemias. Leukemia 2004. 18(4):734-42. (III)


Verpoest et al. The reproductive outcome of female patients with myotonic dystrophy type 1 (DM1) undergoing PGD is not affected by the size of the expanded CTG repeat tract. J Assist Reprod Genet 2010. 27(6):327-33. (III)


Villani et al. Biochemical and imaging surveillance in germline TP53 mutation carriers with Li-Fraumeni syndrome: 11 year follow-up of a prospective observational study. Lancet Oncol 2016. 17(9):1295-305. (III)


Vising et al. Diagnosis of Pompe disease: muscle biopsy vs blood-based assays. JAMA Neurol 2013. 70(7):923-7. (V)


Vladich F. A rapid, automated silver in situ hybridization (SISH) detection assay for HER2 gene status determination in breast carcinoma; 2007. (V)

Voelker. Children's deaths linked with postsurgical codeine. JAMA 2012. 308(10):963. (V)


Vos et al. Opening the psychological black box in genetic counseling. The psychological impact of DNA testing is predicted by the counselee’s perception, the medical impact by the pathogenic or uninformative BRCA1/2-result. Psychooncology 2012. 21(1):29-42. (III)


Wadman et al. Drug treatment for spinal muscular atrophy type I. Cochrane Database Syst Rev 2012. 4:CD006281. (I)


Walerych et al. The rebel angel: mutant p53 as the driving oncogene in breast cancer. Carcinogenesis 2012. (V)


Wei et al. Intratumoral and Intertumoral Genomic Heterogeneity of Multifocal Localized Prostate Cancer Impacts Molecular Classifications and Genomic Prognosticators. Eur Urol 2016. (III)


Wells et al. Revised American Thyroid Association guidelines for the management of medullary thyroid carcinoma. Thyroid 2015. 25(6):567-610. (IV)

Welslau et al. Patient-reported outcomes from EMILIA, a randomized phase 3 study of trastuzumab emtansine (T-DM1) versus capcitabine and lapatinib in human epidermal growth factor receptor 2-positive locally advanced or metastatic breast cancer. Cancer 2014. 120(5):642-51. (II)

Weng and Levy. Genetic polymorphism of the inhibitory IgG Fc receptor FcgammaRIIb is not associated with clinical outcome in patients with follicular lymphoma treated with rituximab. Leukemia & Lymphoma 2009. 50(5):723-7. (V)


Wimmer et al. Spectrum of single- and multiexon NF1 copy number changes in a cohort of 1,100 unselected NF1 patients. Genes Chromosomes Cancer 2006. 45(3):265-76. (III)


Winkel et al. The natural course of non-classic Pompe's disease; a review of 225 published cases. J Neurol 2005. 252(8):875-84 (V)


Winter et al. Assessment of thiopurine methyltransferase enzyme activity is superior to genotype in predicting myelosuppression following azathioprine therapy in patients with inflammatory bowel disease. Aliment Pharmacol Ther 2007. 25(9):1069-77. (II)


Wraith JE. Recommendations on the diagnosis and management of Niemann-Pick disease type C. Mol Genet Metab, 2009. 98(1-2):152-165. (V)
Wu et al. Clinical factors influencing the performance of Gene Expression Classifier testing in indeterminate thyroid nodules. Thyroid 2016. 26(7):916-22. (III)


Wu et al. Melanoma hyperpigmentation is strongly associated with KIT alterations. Am J Dermatopathol 2009. (V)


Xia et al. Frequencies of SF3B1, NOTCH1, MYD88, BIRC3 and IGHV mutations and TP53 disruptions in Chinese with chronic lymphocytic leukemia: disparities with Europeans. Oncotarget 2015. 6(7):5426-34. (III)


Xu et al. HOXB13 is a susceptibility gene for prostate cancer: results from the International Consortium for Prostate Cancer Genetics (ICPCG). Hum Genet 2013. 132(1):5-14. (III)


Yamamoto-Ibusuki et al. Comparison of prognostic values between combined immunohistochemical score of estrogen receptor, progesterone receptor, human epidermal growth factor receptor 2, Ki-67 and the corresponding gene expression score in breast cancer. Mod Pathol 2013. 26(1):79-86. (II)


Yang et al. Promising biomarkers for predicting the outcomes of patients with KRAS wild-type metastatic colorectal cancer treated with anti-epidermal growth factor receptor monoclonal antibodies: a systematic review with meta-analysis. Int J Cancer 2013. 133(8):1914-25. (I)


Yoder et al. Reflex UroVysion testing of bladder cancer surveillance patients with equivocal or negative urine cytology: a prospective study with focus on the natural history of anticipatory positive findings. Am J Clin Pathol 2007. 127(2):295-301. (III)


Yoon et al. Analysis of HER2 status in breast carcinoma by fully automated HER2 fluorescence in situ hybridization (FISH): comparison of two immunohistochemical tests and manual FISH. APMIS 2014. 122(9):755-60. (V)


Yrigollen et al. AGG interruptions and maternal age affect FMR1 CGG repeat allele stability during transmission. J Neurodev Disord 2014. 6(1):24. (III)


Zhang et al. Identification of a distinct mutation spectrum in the SMPD1 gene of Chinese patients with acid sphingomyelinase-deficient Niemann-Pick disease. Orphanet J Rare Dis 2013. 8:15. (V)


Zochling et al. The current concept of spondyloarthritis with special emphasis on undifferentiated spondyloarthritis. Rheumatology (Oxford) 2005. 44(12):1483-91. (V)