Anhang A

Ergänzende Literaturhinweise

Prof. Dr. med. Holger Höhn
Institut für Humangenetik, Universität Würzburg

Außer klassischen Arbeiten berücksichtigt die folgende Auswahl vor allem neuere Publikationen. Ältere Literaturstellen finden sich in den Buchartikeln. Bei mehr als drei Autoren wird aus Platzgründen nur der Erstautor und der Arbeitsgruppenleiter (in Klammern) angegeben, sofern dieser nicht Erstautor ist.

Buchkapitel und Monografien

- Schroeder-Kurth TM, Auerbach AD, Obe G (**1989**) *Fanconi Anemia. Clinical, Cytogenetic and Experimental Aspects.* Berlin, Germany, Springer Verlag
- Alter BP (**1994**) Clinical features of Fanconi anaemia. In: Young NS, Alter BP (eds) *Aplastic Anaemia: Acquired and Inherited*. W.B. Saunders, Philadelphia, pp 275-308
- Auerbach AD, Buchwald M, Joenje H (**2003**) Fanconi anaemia. In: Vogelstein B, Kinzler KW (eds) *The Genetic Basis of Human Cancer*. McGraw-Hill, New York, pp 289-306
- Hoehn H, Thiel-Gross M, Schindler D, et al (**2003**) Genetic Instability and Fanconi Anemia. In: Hisama FM, Weissman SM, Martin GM (eds) *Chromosomal Instability and Aging.* Marcel Dekker, New York and Basel, pp 375-408
- Schindler D, Höhn H (**2003**) *Fanconi Anämie*. medizinischegenetik edition 2, Verlag Medizinische Genetik, München, pp 1-117

Neuere Übersichtsartikel

- Joenje H, Patel KJ (**2001**) The emerging genetic and molecular basis of Fanconi anemia. *Nat Rev Genet* **2**:446-57
- Ahmad SI, Hanaoka F, Kirk SH (**2002**) Molecular biology of Fanconi anaemia – an old problem, a new insight. *Bioessays* **24**:439-48
- Bagby CG Jr (**2003**) Genetic basis of Fanconi anemia. *Curr Opin Hematol* **10**:68-76
- D´Andrea AD, Grompe M (**2003**) The Fanconi anaemia/BRCA pathway. *Nat Rev Cancer* **3**:23-34
- Tischkowitz MD, Hodgson SV (**2003**) Fanconi anaemia. *J Med Genet* **40**:1-10
- Digweed M (**2003**) Response to environmental carcinogens in DNA-repair-deficient disorders. *Toxicology* **193**:111-24

- Venkitaraman AR (**2004**) Tracing the network connecting BRCA and Fanconi anemia proteins. *Nat Rev Cancer* **4**:266-76
- Tischkowitz MD, Dokal I (**2004**) Fanconi anaemia and leukaemia – clinical and molecular aspects. *Br J Haematol* **126**:176-91
- Wang X, D'Andrea AD (**2004**) The interplay of Fanconi anemia proteins in the DNA damage response. *DNA repair (Amst*) **3**:1063-9

Formale Genetik der Fanconi-Anämie (Art der Vererbung)

- Schroeder TM et al (**1976**) Formal genetics of Fanconi´s anemia. *Hum Genet* **32**:257-88
- Rahman N, Ashworth A (**2004**) A new gene on the X involved in Fanconi anemia. *Nat Genet* **36**:1142-3

Krankheitsbild, Krankheitsverlauf (historische Arbeiten)

- Fanconi G (**1927**) Familiäre infantile perniziosaähnliche Anämie (perniziöses Blutbild und Konstitution) *Jahrb Kinderh* **117**:257
- Uehlinger E (**1929**) Konstitutionelle (perniziosaartige) Anämie. *Klin Wochenschr* **32**: 1501
- Gmyrek D, Syllm-Rapoport I (**1964**) Zur Fanconi Anämie (FA). Analyse von 129 beschriebenen Fällen. *Z Kinderheilk* **91**:297
- Fanconi G (**1967**) Familial constitutional panmyelocytopathy, Fanconi´s anemia (F.A.) I. Clinical aspects. *Semin Hematol* **4**:233
- Barmann GJ et al (Opitz JM) (**1977**) Studies of malformation syndromes of man XLVII: disappearance of spermatogonia in the Fanconi anemia syndrome. *Eur J Pediatr* **125**:163-8
- Aynsley-Green A et al (Prader A) (**1978**) Endocrine studies in Fanconi´s anaemia. Report of 4 cases. *Arch Dis Child* **53**:126-31
- Zaizov R, Matoth Y, Mamon Z (**1978**) Longterm observations in children with Fanconi´s anaemia. In: *Aplastic anemia*, pp 243-51
- Chu JY (**1979**) Granulopoiesis in Fanconi´s aplastic anemia. *Proc Soc Exp Biol Med* **161**:609-12
- Glanz A, Fraser FC (**1982**) Spectrum of anomalies in Fanconi anemia. *J Med Genet* **19**:412-420
- Berkovitz GD, Zinkham WH, Migeon CJ (**1984**) Gonadal function in two siblings with Fanconi´s anaemia. *Horm Research* **19**:137-41
- Gastrena J et al (**1986**) Fanconi´s anemia. Clinical study of six cases. *Am J Pediatr Hematol Oncol* **8**:173-7
- Shahidi NT (**1987**) Fanconi anemia, dyskeratosis congenetika, and WT syndrome. *Am J Med Genet Suppl* **3**:263-78
- Rogers PC et al (**1989**) Presentation and outcome of 25 cases of Fanconi´s anemia. *Am J Pediatr Hematol Oncol* **11**:141-5
- Alter BP et al (Auerbach AD) (**1991**) Fanconi´s anaemia and pregnancy. *Br J Haematol* **77**:410-8
- Alter BP et al (**1991**) Erythropoiesis in Fanconi´s anemia. *Blood* **78**:602-8Krankheitsbild, Krankheitsverlauf

Krankheitsbild, Krankheitsverlauf (neuere Beschreibungen und Fallberichte)

- De Kerviler E et al (Gluckman E) (**2000**) The clinical and radiological features of Fanconi´s anaemia. *Clin Radiol* **55**:340-5
- Nowzari H et al **(2001**) Aggressive periodontitis associated with Fanconi´s anemia. A case report. *J Periodontol* **72**:1601-6
- Roxo P Jr et al (**2001**) Allergic and immunologic parameters in patients with Fanconi´s anemia. *Int Arch Allergy Immunol* **125**:349-55
- Olgilvie P, Hofmann UB, Brocker EB, Hamm H (**2002**) Hautveränderungen bei Fanconi Anämie. *Hautarzt* **53**:253-7
- Merriman M, Mora J, McGaughran J (**2002**) Fanconi anemia and primary cataracts: first case. *Ophthalmic Genet* **23**:253-5
- Santos F, Selesnick SH, Glasgold RA (**2002**) Otologic manifestations of Fanconi anemia. *Otol Neurotol* **23**:873-5
- McGaughran J (**2003**) Klippel-Feil anomaly in Fanconi anemia. *Clin Dysmorphol* **12**:197
- Otan F, Acikgoz G, Sakallioglu U, Ozkan B (**2004**) Recurrent apthous ulcers in Fanconi´s anaemia: a case report. *Int J Pediat Dent* **14**:214-7
- Landmann E, Bluetters-Sawatzki R, Schindler D, Gortner L (**2004**) Fanconi anemia in a neonate with pancytopenia. *J Pediatr* **145**:125-7
- Unal S et al (**2004**) Five Fanconi anemia patients with unusual organ pathologies. *Am J Hematol* **77**:50-4
- Aslan D et al (**2005**) An unusual ocular manifestation in Fanconi anemia: Congenital glaucoma. *Am J Hematol* **78**:64-6

Zusammenfassende Arbeiten aus dem Internationalen Fanconi-Anämie-Register (IFAR)

- Auerbach AD, Rogatko A, Schroeder-Kurth TM (**1989**) International Fanconi Anemia Registry: relation of clinical symptoms to diepoxybutane sensitivity. *Blood* **73**:391-6
- Auerbach AD, Allen RG (**1991**) Leukemia and preleukemia in Fanconi anemia patients. A review of the literature and report of the International Fanconi Anemia Registry. *Cancer Genet Cytogenet* **51**:1-12
- Giampietro PF et al (Auerbach AD) (**1993**) The need for more accurate and timely diagnosis in Fanconi anemia: a report from the International Fanconi Anemia Registry. *Pediatrics* **91**:1116-20
- Butturini A et al (Auerbach AD) (**1994**) Hematologic abnormalities in Fanconi anemia: an International Fanconi Anemia Registry study. *Blood* **84**:1650-5
- Wajnrajch MP et al (Auerbach AD) (**2001**) Evaluation of growth and hormonal status in patients referred to the International Fanconi Anemia Registry. *Pediatrics* **107**:744-54
- Kutler DI et al (Auerbach AD) (**2003**) A 20-year perspective on the International Fanconi Anemia Registry (IFAR). *Blood* **101**:1249-56

Fallberichte über erwachsene Patienten mit Fanconi-Anämie

- Liu JM, Auerbach AD, Young NS (**1991**) Fanconi anemia presenting unexpectedly in an adult kindred with no dysmorphic features. *Am J Med* **91**:555-7
- Zatterale A et al (**1995**) Identification and treatment of late onset Fanconi´s anemia. *Haematologica* **80**:535-8
- Rubinstein WS et al (Mulvihill JJ) (**1997**) Interstitial lung disease in an adult with Fanconi anemia. *Am J Med Genet* **69**:315-9
- Kwee ML et al (Joenje H) (**1997**) An atypical case of Fanconi anemia in elderly sibs. *Am J Med Genet* **68;3**:362-6

Differentialdiagnostik der Fanconi-Anämie (FA-ähnliche Syndrome)

- Cox H et al (**1999**) Radial ray defects and associated anomalies. *Clin Genet* **3**5:322-30
- Narchi H (**1999**) Oesophageal atresia, VACTERL association: Fanconi´s anaemia related spectrum of anomalies. *Arch Dis Child* **80**:207
- Temtany SA et al (**2003**) Expanding the phenotypic spectrum of the Baller-Gerold syndrome. *Genet Couns* **14**:299-312
- Bobabilla-Morales L et al (**2003**) Chromosome instability induced in vitro with mitomycin C in five Seckel syndrome patients. *Am J Med Genet* **123**:148-52
- Gennery AR et al (**2004**) The clinical and biological overlap between Nijmegen Breakage Syndrome and Fanconi anemia. *Clin Immunol* **113**:214-9
- New HV et al (**2005**) Nijmegen breakage syndrome diagnosed as Fanconi anemia. *Pediatr Blood Cancer* **44;5**:494-9

Leukämie- und Tumor-Risiko (neuere Übersichtsarbeiten)

- Alter BP (**2003**) Cancer in Fanconi anemia, 1927-2001. *Cancer* **97**:425-40
- Rosenberg PS, Greene MH, Alter BP (**2003**) Cancer incidence in persons with Fanconi anemia. *Blood* **101**:822-6
- Kutler DI et al (**2003**) High incidence of head and neck squamous cell carcinoma in patients with Fanconi anemia. *Achr Otolaryngol Head Neck Surg* **129**:106-12
- Offit K et al (Auerbach AD) (**2003**) Shared genetic susceptibility to breast cancer, brain tumors, and Fanconi anemia. *J Natl Canc Inst* **95**:1548-51
- Kutler DI et al (Auerbach AD) (**2003**) Human papillomavirus DNA and p53 polymorphisms in squamous cell carcinomas from Fanconi anemia patients. *J Natl Cancer Inst* **95**:1718-21
- Rosenberg PS, Huang Y, Alter BP (**2004**) Individualized risks of first adverse events in patients with Fanconi anemia. *Blood* **104**:350-5

Leukämie- und Tumor-Risiko (Fallberichte)

- Swift M et al (**1971**) Squamous cell carcinomas in Fanconi´s anemia. *JAMA* **216**:325-6
- Snow DG et al (Smallman LA) (**1991**) Fanconi´s anemia and post-cricoid carcinoma. *J Laryngol Otol* **105**:125-7
- Lebbe C et al (Morel P) (**1993**) Fanconi´s anaemia associated with multi-centric Bowen´s disease and decreased NK cytotoxicity. *Br J Dermatol* **129**:615
- Alter BP, Tenner MS (**1994**) Brain tumors in patients with Fanconi´s anemia. *Arch Pediatr Adolesc Med* **148**:661-3
- Butturini A et al (Gale RP) (**1994**) Short stature, Fanconi anaemia, and risk of leukemia after growth hormone therapy. *Lancet* **343**:1576
- Lustig JP et al (Sigler E) (**1995**) Head and neck carcinoma in Fanconi´s anaemia - report of a case and review of the literature. *Eur J Cancer B Oral Oncol* **31B**:68-72
- Levinson S, Vincent KA (**1997**) Multifocal osteosarcoma in a patient with Fanconi anemia. *J Pediatr Hematol Oncol* **19**:251-3
- Doerr TD et al (**1998**) Squamous cell carcinoma of the supraglottic larynx in patients with Fanconi´s anemia. *Otolaryngol Head Neck Surg* **118**:523-5
- Goldsby RE et al (Burggers CS) (**1999**) Lymphoblastic lymphoma and excessive toxicity from chemotherapy: an unusual presentation for Fanconi anemia. *J Pediatr Hematol Oncol* **21**:240-3
- Ariffin H et al (**2000**) Wilms tumor and Fanconi anaemia: an unusual association. *J Paediatr Child Health* **36**:196-7
- Ruud E, Wesenberg F (**2001**) Microcephalus, medulloblastoma and excessive toxicity from chemotherapy: an unusual presentation of Fanconi anaemia. *Acta Paediatr* **90**:580-3
- Oksuzoglu B, Yalcin S (**2002**) Squamous cell carcinoma of the tongue in a patient with Fanconi´s anemia: a case report and review of the literature. *Ann Haematol* **81**:294-8
- Bissig H et al (**2002**) Co-occurrence of neuroblastoma and nephroblastoma in an infant with Fanconi´s anemia. *Hum Pathol* **33**:1047-51
- Pavithran K et al (**2002**) Adenocarcinoma of the stomach in Fanconi´s anemia. *Ann Hematol* **81**:666-7
- Tischkowitz MD et al (**2004**) Medulloblastoma as a first presentation of Fanconi anemia. *J Pediatr Hematol Oncol* **26**:52-5
- Harper JL et al (**2004**) Vulvar cancer in a patient with Fanconi´s anemia, treated with 3D conformal radiotherapy. *Am J Hematol* **76**:148-51
- Roginsky R et al (**2004**) Vulvar cancer with Fanconi´s anemia and neutropenic fever: a case report. *J Reprod Med* **49**:218-21

Auffällige Laborbefunde (ohne Chromosomen)

- Froom P et al (Lahat N) (**1987**) Reduced natural killer activity in patients with Fanconi´s anemia and in family members. *Leuk Res* **11**:197-9
- Rosselli F et al (Moustacchi E) (**1992**) Abnormal lymphokine production: a novel feature of the genetic disease Fanconi anemia. I. Involvement of Interleukin-6. *Hum Genet* **89**:42-8

- Schultz JC, Shahidi NT (**1993**) Tumor necrosis factor-alpha overproduction in Fanconi´s anemia. *Am J Hematol* **43**:196-201
- Wunder E et al (Henon PR) (**1993**) Anomalous plasma concentrations and impaired secretion of growth factors in Franconi´s anemia. *Stem Cells (Dayt)* **11**: Suppl 2:144-9
- Bagnara GP et al (**1993**) Production of interleukin 6, leukemia inhibitory factor and GM-CSF by peripheral blood mononuclear cells in Fanconi´s anemia. *Stem Cells (Dayt)* **11**: Suppl 2:137-43
- Roselli et al (Moustacchi E) (**1994**) Abnormal lymphokine production: a novel feature of the genetic disease Fanconi anemia. II. In vitro and in vivo spontaneous overproduction of tumor necrosis factor alpha. *Blood* **83**:1216-25
- Lyman SD et al (Shahidi NT) (**1995**) Plasma/serum levels of flt3 ligand are low in normal individuals and highly elevated in patients with Fanconi anemia and acquired aplastic anemia. *Blood* **86**:4091-6
- Straface E et al (Pagano G) (**2000**) Spectrin changes occur in erythrocytes from patients with Fanconi´s anemia and their parents. *Biochem Biophys Res Commun* **273**:899-01
- Cassinat B et al (Gluckman E) (**2000**) Constitutive elevation of serum alpha-fetoprotein in Fanconi anemia. *Blood* **96**:859-63
- Dupuis-Girod S et al (**2001**) Growth hormone deficiency caused by pituitary stalk interruption in Fanconi´s anemia. *J Pediatr* **138**:129-33
- Dufour C et al (Pistoia V) (**2003**) TNF-alpha and IFN-gamma are overexpressed in the bone marrow of Fanconi anemia patients and TNF-alpha suppresses erythropoiesis in vitro. *Blood* **102**:2053-9

Spontane Chromosomenbrüchigkeit und Veränderungen der Chromosomenenden (Telomere)

- Schroeder TM, Anschütz F, Knopp A (**1964**) Spontane Chromososomenaberrationen bei familiärer Panmyelopathie. *Humangenetik* **1**:194-6
- Schroeder TM, Kurth R (**1971**) Spontaneous chromosomal breakage and high incidence of leukemia in inherited disease. *Blood* **37**:96-112
- Leteurtre F et al (Gluckman E) (**1999**) Accelerated telomere shortening and telomerase activation in Fanconi´s anaemia. *Br J Haematol* **105**:883-93
- Adelfalk C et al (Schweiger M) (**2001**) Accelerated telomere shortening in Fanconi anemia fibroblasts – a longitudinal study. *FEBS Lett* **506**:22-6
- Maluf SW, Erdtmann B (**2001**) Genomic instability in Down syndrome and Fanconi anemia assessed by micronucleus analysis and single-cell gel electrophoresis. *Cancer Genet Cytogenet* **124**:71-5
- Callen E et al (Surralles J) (**2002**) Relationship between chromosome fragility, aneuploidy and severity of the haematological disease in Fanconi anaemia. *Mutat Res* **504**:75-83
- Li X et al (Gluckman E) (**2003**) Abnormal telomere metabolism in Fanconi´s anaemia correlates with genomic instability and the probability of developing severe aplastic anaemia. *Br J Haematol* **120**:836-45

Zelluläre Überempfindlichkeit (Chemikalien)

- Sasaki MS, Tonomura A (**1973**) A high susceptibility of Fanconi´s anemia to chromosome breakage by DNA cross-linking agents. *Cancer Res* **33**:1829
- Auerbach AD, Woman SR (**1976**) Susceptibility of Fanconi´s anaemia fibroblasts to chromosome damage by carcinogens. *Nature* **261**:494-6
- Weksberg R, Buchwald M et al (**1979**) Specific cellular defects in patients with Fanconi anemia. *J Cell Physiol* **101**:311-23
- Kaiser TN et al (**1982**) Flow cytometric characterization of the response of Fanconi´s anemia cells to mitomycin C treatment. *Cytometry* **2**:291-7
- Digweed M, Zakrzewski-Ludcke S, Sperling K (**1988**) Fanconi´s anaemia: correlation of genetic complementation group with psoralen/UVA response. *Hum Genet* **78**:51-4
- Carreau M et al (Buchwald M) (**1999**) Drug sensitivity spectra in Fanconi anemia lymphoblastoid cell lines of defined complementation groups. *Mutat Res* **435**:103-9
- Pagano G, Manini P, Bagchi D (**2003**) Oxidative stress-related mechanisms are associated with xenobiotics exerting excess toxicity to Fanconi anemia cells. *Environ Health Perspect* **111**:1699-703

Zelluläre Überempfindlichkeit (Strahlen)

- Bigelow SB, Rary JM, Bender MA (**1979**) G2 chromosomal radiosensitivity in Fanconi´s anemia. *Mutat Res* **63**:189-99
- Arlett CF, Harcourt SA (**1980**) Survey of radiosensitivity in a variety of human cell strains. *Cancer Res* **40,3**:926-32
- Knox SJ et al (Misra H) (**1981**) Increased radiosensitivity of a subpopulation of T-lymphocyte progenitors from patients with Fanconi´s anemia. *Blood* **57**:1043-8
- Duckworth-Rysiecki G, Taylor AM (**1985**) Effects of ionizing radiation on cells from Fanconi´s anemia patients. *Cancer Res* **45**:416-20
- Gluckman E (**1990**) Radiosensitivity in Fanconi anemia: application to the conditioning for bone marrow transplantation. *Radiother Oncol* **18** S1:88
- Burnet NG et al (Peacock JH) (**1994**) Cellular sensitivity and low dose-rate recovery in Fanconi anaemia fibroblasts. *Br J Radiol* **67**:579-83
- Djuzenova CS et al (**2001**) Response to X-irradiation of Fanconi anemia homozygous and heterozygous cells assessed by the single-cell gel electrophoresis (comet) assay. *Lab Invest* **81**:185-92
- Bremer M et al (**2003**) Fanconi´s anemia and clinical radiosensitivity: report on two adult patients with locally advanced solid tumors treated by radiotherapy. *Strahlenther Onkol* **179**:748-53
- Kalb R et al (Schindler D) (**2004**) Lack of sensitivity of primary Fanconi´s anemia fibroblasts to UV and ionizing radiation. *Radiat Res* **161**:318-25
- Djuzenova C, Flentje M, Plowman PN (**2004**) Radiation response in vitro of fibroblasts from a Fanconi anemia patients with marked clinical radiosensitivity. *Strahlenther Onkol* **180**:789-97

Zelluläre Überempfindlichkeit (Sauerstoff)

- Joenje H, Arwert F et al (**1981**) Oxygen-dependence of chromosomal aberrations in Fanconi´s anaemia. *Nature* **290**:142-3
- Dallapiccola B et al (**1985**) Effects of oxidants and antioxidants on chromosomal breakage in Fanconi anemia lymphocytes. *Hum Genet* **69**:62-5
- Gille JJ, Wortelboer HM, Joenje H (**1987**) Antioxidant status of Fanconi anaemia fibroblasts. *Hum Genet* **77**:28-31
- Schindler D, Hoehn H (**1988**) Fanconi anemia mutation causes cellular susceptibility to ambien oxygen. *Am J Hum Genet* **43**:429-35
- Saito H, Hammond AT, Moses RE (**1993**) Hypersensitivity to oxygen is a uniform and secondary defect in Fanconi anemia cells. *Mutat Res* **294**:255
- Pagano G et al (**1997**) In vitro hypersensitivity to oxygen of Fanconi anemia (FA) cells is linked to ex vivo evidence for oxidative stress in FA homozygotes and heterozygotes. *Blood* **89**:1111-2
- Liebetrau W et al (Hoehn H) (**1997**) Mutagenic activity of ambient oxygen and mitomycin C in Fanconi´s anaemia cells. *Mutagenesis* **12**:69-77
- Ruppitsch W et al (Schweiger M) (**1997**) The role of oxygen metabolism for the pathological pheno-type of Fanconi anemia. *Hum Genet* **99**:710-19
- Will O, Schindler D, Boiteux S, Epe B (**1998**) Fanconi´s anaemia cells have normal steady-state levels and repair of oxidative DNA base modifications sensitive to Fpg protein. *Mutat Res* **409**:65-72
- Pagano G (**2000**) Mitomycin C and diepoxybutane action mechanisms and FANCC protein functions: further insights into the role of oxidative stress in Fanconi´s anaemia phenotype. *Carcinogenesis* **21:**1067-8
- Pagano G, Youssoufian H (**2003**) Fanconi anemia proteins: majors roles in cell protection against oxidative damage. *Bioessays* **25**:589-95
- Park SJ et al (Clapp DW) (**2004**) Oxidative stress/damage induced multimerization and interaction of Fanconi anemia proteins. *J Biol Chem* **279**:30053-9
- Pagano G et al (**2004**) Gender- and age-related distinctions for the in vivo prooxidant state in Fanconi anaemia patients. *Carcinogenesis* **25**:1899-909

Zellwachstum und Zellzyklus

- Elmore E, Swift M (**1975**) Growth of cultured cells from patients with Fanconi anemia. *J Cell Physiol* **87**:229-33
- Dutrillaux B et al (**1981**) The cell cycle of lymphocytes in Fanconi anemia. *Hum Genet* **62**:327-32
- Kubbies M et al (Hoehn H) (**1985**) Endogenous blockage and delay of the chromosome cycle in-spite normal recruitment and growth phase explain poor proliferation and frequent endomitosis in Fanconi anemia cells. *Am J Hum Genet* **37**:1022-29
- Seyschab H et al (Hoehn H) (**1993**) G2 phase cell cycle disturbance as a manifestation of genetic cell damage. *Hum Genet* **92**:61-68
- Poot M et al (Hoehn H) (**1996**) Cell cycle defect in connection with oxygen and iron sensitivity in Fanconi anemia lymphoblastoid cells. *Exp Cell Res* **222**:262-8

- Heinrich MC et al (Bagby GC) (**1998**) DNA cross-linker induced G2/M arrest in group C Fanconi anemia lymphoblasts reflects normal checkpoint function. *Blood* **91**:275-87
- Sala-Trepat M et al (Papadopoulo D) (**2000**) Arrest of S-phase progression is impaired in Fanconi anemia cells. *Exp Cell Res* **260**:208-15
- Akkari YM et al (Grompe M) (**2001**) The 4N cell cycle delay in Fanconi anemia reflects growth arrest in late S phase. *Mol Genet Metab* **74**:403-12
- Li X, et al (Haneline LS) (**2003**) Fanconi anemia type C-deficient hematopoietic stem/progenitor cells exhibit aberrant cell cycle control. *Blood* **102**:2081-4
- Freie BW et al (Clapp DW) (**2004**) A role for the Fanconi anemia C protein in maintaining the DNA damage-induced G2 checkpoint. *J Biol Chem* **279**:50986-93

Hinweise auf DNA-Reparaturdefekte

- Lambert MW et al (Parrish DD) (**1992**) Defective DNA endonuclease activities in Fanconi´s anemia cells, complementation groups A and B. *Mutat Res* **273**:57-71
- Guillof C et al (Papadopoulo D) (**1993**) Mutagenic processing of psoralen monoadducts differ in normal and Fanconi anemia cells. *Mutagenesis* **8**:355-61
- Zhen W et al (Bohr VA) (**1993**) Deficient gene specific respair of cisplatin-induced lesions in Xeroderma pigmentosum and Fanconi´s anemia cell lines. *Carcinogenesis* **14**:919-24
- Laguerbe A et al (Papadopoulo D) (**1995**) The molecular mechanism underlying formation of deletions in Fanconi anemia cells may involve site-specific recombination. *Proc Natl Acad Sci (USA)* **92**:831-5
- Thyagarajan B, Campell C (**1997**) Elevated homologous recombination activity in fanconi anemia fibroblasts. *J Biol Chem* **272**:23328-33
- Smith J et al (Papadopoulo D) (**1998**) Abnormal rearrangements associated with V(D)J recombination in Fanconi anemia. *J Mol Biol* **281**:815-25
- Buchwald M, Moustacchi E (**1998**) Is Fanconi anemia caused by a defect in the processing of DNA damage? *Mutat Res* **408**:75-90
- Lambert MW, Lambert WC (**1999**) DNA repair and chromatin structure in genetic diseases. *Prog Nucl Acid Res Mol Biol* **63**:257-310
- Digweed M et al (**2002**) Attenuation of the formation of DNA-repair foci containing RAD51 in Fanconi anaemia. *Carcinogenesis* **23**:1121-6
- Ramirez MH et al (Schweiger M) (**2003**) The cellular control enzyme polyADP ribosyl transferase is eliminated in cultured Fanconi anemia fibroblasts at confluency. *Biol Chem* **384**:169-74
- Donahue SL et al (**2003**) Deficient regulation of DNA double-strand break repair in Fanconi anemia fibroblasts. *J Biol Chem* **278**:29487-95
- Donahue SL, Campbell C (**2004**) A Rad50-dependent pathway of DNA repair is deficient in Fanconi anemia fibroblasts. *Nucleic Acids Res* **32**:3248-57

Labordiagnostik, prä- und postnatal (Zytogenetik, Zellzyklus)

- Auerbach AD, Adler B, Chaganti RSK (**1981**) Prenatal and postnatal diagnosis and carrier detection of Fanconi anemia by a cytogenetic method. *Pediatrics* **67**:128-35
- Schindler D et al (**1985**) Presymptomatic diagnosis of Fanconi´s anemia. *Lancet* **1**:937
- Miglierina R, Le Coniat M, Berger R (**1991**) A simple diagnostic test for Fanconi anemia by flow-cytometry. *Anal Cell Pathol* **3**:111-114
- Auerbach AD (**1993**) Fanconi anemia diagnosis and the diepoxybutane (DEB) test. *Exp Hematol* **21**:731
- Seyschab H et al (Hoehn H) (**1995**) Comparative Evaluation of Diepoxybutane Sensitivity and Cell Cycle Blockage in the Diagnosis of Fanconi Anemia. *Blood* **85**:2233-7
- Joenje H et al (**1999**) Confounding factors in the diagnosis of Fanconi anaemia. *Am J Med Genet* **79**:403-5
- Fabio T, Crescenzio N, Saracco P (**2000**) Cell cycle analysis in the diagnosis of Fanconi´s anemia. *Haematologica* **85**:431-2
- Terclani S et al (Holzgreve W) (**2001**) Fanconi anemia associated with increased nuchal translucency detected by first-trimester ultrasound. *Ultrasound Obstet Gynecol* **17**:160-2
- Pearson T et al (Joubert G) (**2001**) Fanconi anemia. A statistical evaluation of cytogenetic results obtained from South African families. *Cancer Genet Cytogenet* **126**:52-5
- Esmer C et al (Carnevale A) (**2004**) DEB test for Fanconi anemia detection in patients with atypical phenotypes. *Am J Med Genet* **124**:35-9

Labordiagnostik (FANCD2-Westernblot)

- Shimamura A et al (d´Andrea AD) (**2002**) A novel diagnostic screen for defects in the Fanconi anemia pathway. *Blood* **100**:4649-54
- Shimamura A, d´Andrea AD (**2003**) Subtyping of Fanconi anemia patients: implications for clinical management. *Blood* **102**:4359

Präimplantationsdiagnostik

- Josefson D (**2000**) Couple select healthy embryo to provide stem cells for sister. *BMJ* **321**:917
- Verlinsky Y et al (Kuliev A) (**2001**) Preimplantation diagosis for Fanconi anemia combined with HLA matching. *JAMA* **285**:3130-3
- Damewood MD (**2001**) Ethical implications of a new application of preimplantation diagnosis. *JAMA* **285**:3143-4
- Wolf SM, Kahn JP, Wagner JE (**2003**) Using preimplantation genetic diagnosis to create a stem cell donor: issues, guidelines and limits. *J Law Med Ethics* **31**:327-39

- Bielorai B et al (**2004**) Successful umbilical cord transplantation for Fanconi anemia using preimplantation genetic diagnosis for HLA-matched donor. *Am J Hematol* **77**:397-9

Klonale Chromosomenaberrationen in Blut und Knochenmark

- Alter BP et al (**1993**) Clonal chromosomal abnormalities in Fanconi´s anaemia: what do they really mean? *Br J Haematol* **85**:627-30
- Berger R et al (**1993**) Chromosome abnormalities in bone marrow of Fanconi anemia patients. *Cancer Genet Cytogenet* **65**:47-50
- Berger R, Jonveaux P (**1996**) Clonal chromsome abnormalities in Fanconi anemia. *Hematol Cell Ther* **38**:291-6
- Ferro MT et al (**2001**) Triplication of 1q in Fanconi anemia. *Cancer Genet Cytogenet* **127**:38-41
- Oliviera NI et al (**2001**) Two different karyotypes with 1q abnormalities in a patient with Fanconi anemia. *Leuk Res* **26**:1047-9
- Tönnies H et al (Neitzel H) (**2003**) Clonal chromosomal aberrations in bone marrow cells of Fanconi anemia patients: gains of the chromosomal segment 3q26q29 as an adverse risk factor. *Blood* **101**:3872-4

Mosaik-Bildung bei Fanconi-Anämie (somatische Reversion)

- Schroeder TM et al (**1977**) Clinical and cytogenetic observations during a six-year period in an adult with Fanconi´s anemia. *Blut* **34**:119-32
- Schroeder TM et al (**1979**) Fanconi´s anemia: terminal leukemia and „Forme fruste“ in one family. *Clin Genet* **16**:260-8
- Kwee ML et al (Joenje H) (**1983**) Unusual response to bifunctional alkylating agents in a case of Fanconi anaemia. *Hum Genet* **64**:384-7
- Dokal I et al (**1996**) Positive diepoxybutane test in only one of two brothers found to be compound heterozygotes for Fanconi´s anaemia complementation group C mutations. *Br J Haematol* **93**:813-6
- Lo Ten Foe JR et al (Joenje H) (**1997**) Somatic mosaicism in Fanconi anemia: molecular basis and clinical significance. *Eur J Hum Genet* **5**:137-48
- Waisfisz Q et al (Joenje H) (**1999**) Sponaneous functional correction of homozygous Fanconi anaemia allels reveals novel mchanistic basis for reverse mosaicism. *Nature Genet* **22**:379-83
- Gregory JJ Jr et al (Auerbach AD) (**2001**) Somatic mosaicism in Fanconi anemia: evidence of genotypic reversion in lymphohematopoietic stem cells. *Proc Natl Acad Sci USA* **98**:2532-7
- Gross M et al (Hoehn H) (**2002**) Reverse mosaicism in Fanconi anemia: natural gene therapy via molecular self-correction. *Cytogenet Genome Res* **98**:126-35
- Hirschhorn R (**2003**) In vivo reversion to normal of inherited mutations in humans. *J Med Genet* **40**:721-8
- Ikeda H et al (Joenje H) (**2003**) Genetic reversion in an acute myelogenous leukemia cell line from a Fanconi anemia patient with biallelic mutations in BRCA2. *Cancer Res* **63**:2688-94

- Soulier J et al (Gluckman E) (**2005**) Detection of somatic mosaicism and classification of Fanconi anemia patients by analysis of the FA/BRCA pathway. *Blood* **105;3**:1329-36

Komplementationsgruppen

- Zakrzewski S, Sperling K (**1980**) Genetic heterogeneity of Fanconi´s anemia demonstrated by somatic cell hybrids. *Hum Genet* **56**:81-4
- Zakrzewski S, Koch M, Sperling K (**1983**) Complementation studies between Fanconi´s anemia cells with different DNA repair characteristics. *Hum Genet* **64**:55-7
- Strathdee CA, Duncan AMW, Buchwald M (**1992**) Evidence for at least four Fanconi anemia genes including FACC on chromosome 9. *Nature Genet* **1**:196-200
- Joenje H et al (**1995**) Fanconi anemia complementation groups in Germany and the Netherlands. *Hum Genet* **97**:280-82
- Joenje H et al (**1995**) Classification of Fanconi anemia patients by complementation analysis: evidence for a fifth genetic subtype. *Blood* **86**:2156
- Joenje H et al (**1997**) Evidence for at least eight Fanconi anemia genes. *Am J Hum Genet* **61**:940-4
- Joenje H et al (**2000**) Complementation analysis in Fanconi anemia: assignement of the reference FA-H patient to group A. *Am J Hum Genet* **67**:759-62
- Hanenberg H et al (**2002**) Phenotypic correction of primary Fanconi anemia T cells with retroviral vectors as a diagnostic tool. *Exp Hematol* **30**:410-20
- Levitus M et al (Joenje H) (**2004**) Heterogeneity in Fanconi anemia: evidence for 2 new genetic subtypes. *Blood* **103**:2498-503

Fanconi-Anämie-Gene: A-Gen (FA-A; *FANCA*)

- Pronk JC et al (Joenje H) (**1995**) Localisation of the Fanconi anaemia complementation group A gene to chromosome 16q24.3. *Nature Genet* **11**:338-40
- Lo Ten Foe JR (Joenje H) (**1996**) Expression cloning of a cDNA for the major Fanconi anaemia gene, FAA. *Nature Genet* **14**:320-3
- Ianzano L et al (Savoia A) (**1997**) The genomic organisation of the Fanconi anemia group A gene. *Genomics* **41**:309-14
- Levran O et al (Auerbach AD) (**1997**) Sequence variation in the Fanconi anemia gene FAA. *Proc Natl Acad Sci (USA)* **94**:13051-6
- Centra M et al (Savoia A) (**1998**) Fine exon-intron structure of the Fanconi anemia group A gene and characterization of two genomic deletions. *Genomics* **51**:463-7
- Levran O, Doggett NA, Auerbach AD (**1998**) Identification of Alu-mediated deletions in the Fanconi anemia gene FAA. *Hum Mutat* **12**:145-53

- Wijker M et al (Mathew C) (**1999**) Heterogeneous spectrum of mutations in the Fanconi anaemia group A gene. *Eur J Hum Genet* **7**:52-9
- Morgan NV et al (Mathew CG) (**1999**) High frequency of large intragenic deletions in the Fanconi group A gene. *Am J Hum Genet* **65**:1330
- Tipping AJ et al (Mathew CG) (**2001**) Molecular and genealogical evidence for a founder effect in Fanconi anemia families of the Afrikaner population of South Africa. *Proc Natl Acad Sci USA* **98**:5734-9
- Savino M et al (Savoia A) (**2003**) Spectrum of FANCA mutations in Italian Fanconi anemia patients. *Hum Mutat* **22**:338-9
- Callen E et al (Surralles J) (**2003**) Quantitative PCR analysis reveals a high incidence of large intragenic deletions in the FANCA gene in Spanish Fanconi anemia patients. *Cytogenet Genome Res* **104**:341-5
- Callen E et al (Surralles J) (**2005**) A common founder mutations in FANCA underlies the world highest prevalence of Fanconi anemia in gypsy families from Spain. *Blood* **105;5**:1946-9

Fanconi-Anämie-Gene: B-Gen (FA-B; *FANCB*)

- Meetei AR, Levitus M, Xue Y et al (Joenje H) (**2004**) X-linked inheritance of Fanconi anemia complementation group B. *Nat Genet* **36**:1219-24
- Fei P, Yin J, Wang W (**2005**) New advances in the DNA damage response network of Fanconi anemia and BRCA proteins: FAAP95 replaces BRCA2 as the true FANCB protein. *Cell Cycle* **4;1**:80-6

Fanconi-Anämie-Gene: C-Gen (FA-C; FACC; *FANCC*)

- Strathdee CA et al (Buchwald M) (**1992**) Cloning of cDNAs for Fanconi´s anaemia by functional complementation. *Nature* **356**:763-7
- Gibson RA et al (Mathew CG) (**1993**) Characterization of the exon structure of the Fanconi anaemia group C gene by vectorette PCR. *Hum Mol Genet* **2**:35-8
- Verlander PC et al (Auerbach AD) (**1995**) Mutation analysis of the Fanconi anemia gene FACC. *Am J Hum Genet* **54**:595-601
- Gibson RA et al (Mathew CG) (**1996**) Novel mutations and polymorphisms in the Fanconi anemia group C gene. *Hum Mutat* **8**:140-8
- Krasnoshtein F, Buchwald M (**1996**) Developmental expression of the Fac gene correlates with congenital defects in Fanconi anemia patients. *Hum Mol Genet* **5**:85-93
- Escarceller M et al (Papadopoulo D) (**1998**) Fanconi anemia C gene product plays a role in the fidelity of blunt end DNA-joining. *J Mol Biol* **279;2**:375-85
- Lackinger D et al (Schweiger M) (**1998**) Involvement of the Fanconi proteine FA-C in repair processes of oxidative DNA damages. *FEBS letters* **440**:103-6
- Haneline LS et al (Buchwald M) (**1999**) Loss of FancC function results in decreased hematopoietic stem cell repopulating ability. *Blood* **94**:1-8

- Cumming RC et al (Buchwald M) (**2001**) Fanconi anemia group C protein prevents apoptosis in hematopoietic cells through redox regulation of GSTP1. *Nat Med* **7**:814-20
- Pang Q et al (Bagby GC) (**200**1) The Fanconi anemia complementation group C gene product: structural evidence of multifunctionality. *Blood* **98**:1392-401
- Freie B et al (Clapp DW) (**2003**) Fanconi anemia type C and p53 cooperate in apoptosis and tumorigenesis. *Blood* **102**:4146-52
- Zanier R et al (Rosselli F) (**2004**) Fanconi anemia C gene product regulates expression of genes involved in differentation and inflammation. *Oncogene* **23**:50004-13
- Niedzwiedz W et al (Patel KJ) (**2004**) The Fanconi anemia gene FANCC promotes homologous recombination and error-prone DNA repair. *Mol Cell* **15**:607-20

Fanconi-Anämie-Gene: D1-Gen (FA-D1; *FANCD1/BRCA2*)

- Howlett NG et al (d´Andrea AD) (**2002**) Biallelic inactivation of BRCA2 in Fanconi anemia. *Science* **297**:606-9
- Popp H et al (Schindler D) (**2003**) Screening Fanconi anemia lymphoid cell lines of non-A, C, D2, E, F, G subtypes for defects in BRCA2/ FANCD1. *Cytogenet Genome Res* **103**:54-7
- Wagner JE et al (**2004**) Germline mutations in BRCA2: shared genetic susceptibility to breast cancer, early onset leukemia, and Fanconi anemia. *Blood* **103**:3226-9
- Hirsch B et al (d'Andrea AD) (**2004**) Association of biallelic BRCA2/ FANCD1 mutations with spontaneous chromosomal instability and solid tumors of childhood. *Blood* **103;7**:2554-9

Fanconi-Anämie-Gene: D2-Gen (FA-D2; *FANCD2*)

- Whitney M et al (Grompe M) (**1995**) Microcell mediated chromosome transfer maps the Fanconi anaemia group D gene to chromosome 3p. *Nature Genet* **11**:341-3
- Hejna JA et al (Grompe M) (**2000**) Localization of the Fanconi anemia complementation group D gene to a 200-kb region on chromosome 3p25.3. *Am J Hum Genet* **66**:1540-51
- Timmers C, Taniguchi T, Hejna J et al (Grompe M) (**2001**) Positional cloning of a novel Fanconi anemia gene, FANCD2. *Mol Cell* **7**:241-8
- Taniguchi T et al (d´Andrea AD) (**2002**) Convergence of the Fanconi anemia and ataxia telangiectasia signaling pathways. *Cell* **109**:459-72
- Nakanishi K et al (d´Andrea AD) (**2002**) Interaction of FANCD2 and NBS1 in the DNA damage response. *Nat Cell Biol* **4**:913-20
- Holzel M et al (de Winter JP) (**2003**) FANCD2 protein is expressed in proliferating cells of human tissues that are cancer-prone in Fanconi anaemia. *J Pathol* **201**:198-203

- Hussain S et al (Mathew CG) (**2004**) Direct interaction of FANCD2 with BRCA2 in DNA damage response pathways. *Hum Mol Genet* **13**:1241-8
- Montes de Oca R, Grompe M et al (d'Andrea AD) (**2005**) Regulated interaction of the Fanconi Anemia protein, FANCD2, with chromatin. *Blood* **105;3**:1003-9

Fanconi-Anämie-Gene: E-Gen (FA-E; *FANCE*)

- Wegner RD, Henrichs I, Joenje H, Schroeder-Kurth TM (**1996**) Fanconi anemia complementation group E: clinical and cytogenetic data of the first patient. *Clin Genet* **50**:479-82
- Waisfisz Q et al (Digweed M) (**1999**) The Fanconi anemia group E gene, FANCE, maps to chromosome 6p. *Am J Hum Genet* **64**:1400-5
- de Winter JP et al (Joenje H) (**2000**) Isolation of a cDNA representing the Fanconi anemia complementation group E gene. *Am J Hum Genet* **67**:1306-8
- Pace P et al (Patel KJ) (**2002**) FANCE: the link between Fanconi anaemia complex assembly and activity. *EMBO J* **21**:3414-23
- Taniguchi T, D'Andrea AD (**2002**) The Fanconi anemia protein FANCE promotes nuclear accumulation of FANCC. *Blood* **100**:2457-62

Fanconi-Anämie-Gene: F-Gen (FA-F; *FANCF*)

- de Winter JP et al (Joenje H) (**2000**) The Fanconi anemia gene FANCF encodes a novel protein with homology to ROM. *Nat Genet* **24**:15-6
- Leveille F et al (Joenje H) (**2004**) The Fanconi anemia gene product FANCF is a flexible adaptor protein. *J Biol Chem* **279**:39421-30

Fanconi-Anämie-Gene: G-Gen (FA-G, *FANCG*/*XRCC9*)

- Saar K et al (Digweed M) (**1998**) Localisation of a Fanconi anaemia gene to chromosome 9p. *Eur J Hum Genet* **6**:501-8
- de Winter JP et al (Joenje H) (**1998**) The Fanconi anaemia group G gene FANCG is identical with XRCC9. *Nature Genet* **20**:281-3
- Demuth I et al (Digweed M) (**2000**) Spectrum of mutations in the Fanconi anaemia group G gene, FANCG/XRCC9. *Eur J Hum Genet* **8**:861-8
- Auerbach AD et al (**2003**) Spectrum of sequence variation in the FANCG gene: an International Fanconi Anemia Registry (IFAR) study. *Hum Mutat* **22**:158-68
- Hussain S et al (Mathew CG) (**2003**) Direct interaction of the Fanconi anaemia protein FANCG with BRCA2/FANCD1. *Hum Mol Genet* **12**:2503-10
- Blom E et al (Joenje H) (**2004**) Multiple TPR motify charaterize the Fanconi anemia FANCG protein. *DNA Repair (Amst)* **3**:77-84

Fanconi-Anämie-Gene: L-Gen (FA-L; *FANCL)*

- Meetei AR et al (Wang W) (**2003**) A novel ubiquitn ligase is deficient in Fanconi anemia. *Nat Genet* **35**:165-70
- Meetei AR, Yan Z, Wang W (**2004**) FANCL replaces BRCA1 as the likely ubiquitin ligase responsible for FANCD2 monoubiquination. *Cell Cycle* **3**:179-81

Genotyp-Phänotyp-Korrelationen

- Yamashita T et al (D´Andrea AD) (**1996**) Clinical variability of Fanconi anemia (type C) results from expression of an amino terminal truncated Fanconi anemia complementation group C polypeptide with partial activity. *Blood* **87**:4424-32
- Gillio AP et al (Auerbach AD) (**1997**) Phenotypic consequences of mutations in the Fanconi anemia FAC gene: an International Fanconi Anemia Registry study. *Blood* **90**:105-10.
- Koc A et al (Altay C) (**1999**) Variable pathogenicity of exon 43del (FAA) in four Fanconi anaemia patients within a consanguineous family. *Br J Haematol* **104**:127-30.
- Faivre L et al (Mathew CG) (**2000**) Association of complementation group and mutation type with clinical outcome in Fanconi anemia. European Fanconi Anemia Research Group. *Blood* **96**:4064-70

Tiermodelle für die Fanconi-Anämie

- Chen M et al (Buchwald M) (**1996**) Inactivation of Fac in mice produces inducible chromosomal instability and reduced fertility reminiscent of Fanconi anaemia. *Nature Genet* **12**:448-51
- Whitney MA et al (Grompe M) (**1996**) Germ cell defects and hematopoietic hypersensitivity to gamma-interferon in mice with targeted disruption of the Fanconi anemia C gene. *Blood* **88**:49-58
- Carreau M et al (Buchwald M) (**1998**) Bone marrow failure in the Fanconi anemia group C mouse model after DNA damage. *Blood* **91**:2737
- Battaile KP et al (Grompe M) (**1999**) In vivo selection of wild-type hematopoietic stem cells in a murine model of Fanconi anemia. *Blood* **94**:2151-8
- Otsuki T et al (Liu JM) (**1999**) Tumor necrosis factor-alpha and CD95 ligation suppress erythropoiesis in Fanconi anemia C gene knockout mice. *J Cell Physiol* **189**:79-86
- Cheng NC et al (Arwert F) (**2000**) Mice with targeted disruption of the Fanconi anemia homolog Fanca. *Hum Mol Genet* **9**:1805-11
- Hadjur S et al (Buchwald M) (**2001**) Defective hematopoiesis and hepatic steatosis in mice with combined deficiencies of the genes encoding Fancc and Cu/Zn superoxide dismutase. *Blood* **98**:1003-11
- Yang Y et al (d´Andrea AD) (**2001**) Targeted disruption of the murine Fanconi anemia gene Fancg/Xrcc9. *Blood* **98**:3435-40

- Koomen M et al (Arwert F) (**2002**) Reduced fertility and hypersensitivity to mitomycin C characterize Fancg/Xrcc9 null mice. *Hum Molec Genet* **11**:273-81
- Noll M et al (Grompe M) (**2002**) Fanconi anemia group A and C double-mutant mice: functional evidence for a multi-protein Fanconi anemia complex. *Exp Hematol* **30**:679-88
- Wong JC et al (Buchwald M) (**2003**) Targeted disruption of exons 1 and 6 of the Fanconi anemia group A gene leads to growth retardation, strain-specific microphthalmia, meiotic defects and primordial germ cell hypoplasia. *Hum Mol Genet* **12**:2063-76
- Houghtaling S et al (Grompe M) (**2003**) Epithelial cancer in Fanconi anemia complementation group D2 (Fancd2) knockout mice. *Genes Dev* **17**:2021-35
- Liu TX et al (Look AT) (**2003**) Knockdown of zebrafish Fancd2 causes developmental abnormalities via p53-dependent apoptosis. *Dev Cell* **5**:903-14

Fanconi-Anämie Multi-Protein-Komplex („Core-Komplex“)

- Kupfer GM et al (D´Andrea AD) (**1997**) The Fanconi anaemia proteins, FAA and FAC, interact to form a nuclear complex. *Nature Genet* **17**:487-90
- Yamashita T et al (D´Andrea AD) (**1998**) The fanconi anemia pathway requires FAA phosporylation and FAA/FAC nuclear accumulation. *Proc Natl Acad Sci (USA)* **95**:13085-90
- Waisfisz Q et al (Joenje H) (**1999**) A physical complex of the Fanconi anemia proteins FANCG/XRCC9 and FANCA. *Proc Natl Acad Sci (USA)* **96**:10320-5
- Garcia-Higuera I et al (D´Andrea AD) (**1999**) Fanconi anemia proteins FANCA, FANCC and FANCG/XRCC9 interact in a functional nuclear complex. *Mol Cell Biol* **19**:4866-71
- Reuter T et al (Hoehn H) (**2000**) Strong FANCA-FANCG but weak FANCA-FANCC interaction in the yeast two hybrid system. *Blood* **95;2**:719-20
- Medhurst AL et al (Mathew CG) (**2001**) Direct interactions of the five known Fanconi anaemia proteins suggest a common functional pathway. *Hum Mol Genet* **10**:423-9
- Garcia-Higuera I et al (d'Andrea AD) (**2001**) Interaction of the Fanconi anemia proteins and BRCA1 in a common pathway. *Mol Cell* **7**:249-62
- Gordon SM, Buchwald M (**2003**) Fanconi anemia protein complex: mapping protein interactions in the yeast 2- and 3-hybrid systems. *Blood* **102**:136-41
- Thomashevski A et al (Kupfer GM) (**2004**) The Fanconi anemia core complex forms four complexes of different sizes in different subcellular compartments. *J Biol Chem* **279**:26201-9
- Mi J, Kupfer GM (**2005**) The Fanconi anemia core complex associates with chromatin during S-phase. *Blood* **105;2**:759-66

Zusätzliche Hinweise auf die Funktionen der FA-Gene

- Segal GM et al (Bagby GC) (**1994**) Repression of Fanconi anemia gene (FACC) expression inhibits growth of hematopoietic progenitor cells. *J Clin Invest* **94**:846-52
- Rathbun RK et al (Bagby GC) (**1997**) Inactivation of the Fanconi anemia group C gene augments interferon-gamma-induced apoptotic responses in hematopoietic cells. *Blood* **90**:974-85
- Wang J et al (Liu JM) (**1998**) Overexpression of the fanconi anemia group C gene (FAC) protects hematopoietic progenitors from death induced by Fas-mediated apoptosis. *Cancer Res* **58**:3538-41
- Guillof C et al (Rosselli F) (**1999**) Fanconi anemia C protein acts at a switch between apoptosis and necrosis in mitomycin C-induced cell death. *Exp Cell Res* **246**:384-94
- Qiao F, Moss A, Kupfer GM (**2001**) Fanconi anemia proteins localize to chromatin and the nuclear matrix in a DNA damage- and cell cycle-regulated manner. *J Biol Chem* **276**:23391-6
- Blom E et al (Joenje H) (**2002**) Evolutionary clues to the molecular function of Fanconi anemia genes. *Acta Haematol* **108**:231-6
- Reuter TY et al (**2003**) Yeast two-hyrid screens imply involvement of Fanconi anemia proteins in transcription regulation, cell signaling, oxidative metabolism, and cellular transport. *Exp Cell Res* **289**:211-21

Gentransfer und Gentherapie

- Walsh CE et al (Liu JM) (**1994**) A functionally active retrovirus vector for gene therapy in Fanconi anemia group C. *Blood* **84**:453-9
- Walsh CE et al (Liu JM) (**1995**) Transduction of CD-34 enriched human peripheral and umbilical cord blood progenitors using a retroviral vector with the Fanconi anemia group C gene. *J Investig Med* **43**:379-85
- Fu KL et al (Walsh CE) (**1997**) Functional correction of Fanconi anemia group A hematopoietic cells by retroviral gene transfer. *Blood* **90**:3296-303
- Pulsipher M et al (D´Andrea AD) (**1998**) Subtyping analysis of Fanconi anemia by immunoblotting and retroviral gene transfer. *Mol Med* **4**:468-79
- Liu JM (**1998**) Gene transfer for the eventual treatment of Fanconi´s anemia. *Semin Hematol* **35**:168-79
- Youssoufian H et al (**1998**) Protein replacement by receptor-mediated endocytosis corrects the sensitivity of Fanconi anemia group C cells to mitomycin C. *Blood* **93**:363-9
- Liu JM et al (Walsh CE) (**1999**) Engraftment of hematopoietic progenitor cells transduced with the Fanconi anemia group C gene. *Hum Gene Ther* **10**:2337-46
- Dirksen U et al (Hanenberg H) (**1999**) Fanconi anemia and beta c deficiency-associated pulmonary alveolar proteinosis as two hereditary diseases of childhood which are potentially curable by stem cell gene therapy but require different therapeutic approaches. *Klin Pediatr* **211**:329-35

- Noll M et al (Grompe M) (**2001**) Preclinical protocol for in vivo selection of hematopoietic stem cells corrected by gene therapy in Fanconi anemia group C. *Mol Ther* **3**:14-23
- Yamada K et al (Walsh CE) (**2001**) Functional correction of Fanconi anemia group C hematopoietic cells by the use of a novel lentiviral vector. *Mol Ther* **3**:485-90
- Galimi F et al (Grompe M) (**2002**) Gene therapy in Fanconi anemia: preclinical efficacy using lentiviral vectors. *Blood* **100**:2732-6
- Croop JM (**2003**) Gene therapy for Fanconi anemia. *Curr Hematol rep* **2**:335-40
- Yamada K et al (Walsh CE) (**2003**) Phenotype correction of Fanconi anemia group A hematopoietic stem cells using lentiviral vector. *Mol Ther* **8**: 600-10
- Ferrer M et al (Kruyt FA) (**2004**) Chemosensitizing tumor cells by targeting the Fanconi anemia pathway with an adenovirus overexpressing dominant-negative FANCA. *Cancer Gene Ther* **11**:539-46

Konventionelle Therapie: Androgene

- Shahidi NT (**1973**) Androgens and erythropoiesis. *N Engl J Med* **289**:72-80
- Shahidi NT (**1978**) Androgen therapy in children with aplastic anemia. In: *Aplastic anemia*, pp 409-413
- Molinari PF (**1982**) Erythropoietic mechanism of androgens: a critical review and clinical implications. *Haematologica* **67**:442-60
- Hinterberger W, Vierhapper H (**1993**) Anabolic steroids and blood cell production. *Wien Med Wochenschr* **143**:380-2

Lebertumoren bei Androgen-Behandlung

- Mulvihill JJ et al (**1971**) Hepatic adenoma in Fanconi anemia treated with oxymetholone. *J Pediatr* **87**:122-4
- LeBrun DP et al (Phillips MJ) (**1991**) Fibrolamellar carcinoma of the liver in a patient with Fanconi anemia. *Hum Pathol* **22**:396-8
- Touraine RL et al (Phillippe N) (**1993**) Hepatic tumors during androgen therapy in Fanconi anaemia. *Eur J Pediatr* **152**:691-3
- Resnick MB et al (**1995**) Hepatic adenoma in pediatric age group. *Am J Surg Path* **19**:1181-90
- Kulmar AR, Wagner JE, Auerbach AD et al (**2004**) Fatal hemorrhage from androgen-related hepatic adenoma after hematopoietic cell transplantation. *J Pediatr Hematol Oncol* **26**:16-8
- Velazquez I, Alter BP (**2004**) Androgens and liver tumors: Fanconi´s anemia and non-Fanconi´s conditions. *Am J Hematol* **77**:257-67

Konventionelle Therapie: Zytokine

- Kemahli S et al (Arcasoy A) (**1994**) GM-CSF in the treatment of Fanconi´s anaemia. *Br J Haematol* **87**:871-2

- Guinan EC et al (Nathan DG) (**1994**) Evaluation of granulocyte-macrophage colony-stimulating factor for the treatment of pancytopenia in children with fanconi anemia. *J Pediatr* **124**:144-50
- Rackoff WR et al (Willisams DA) (**1996**) Prolonged administration of granulocyte colony-stimulating factor (filgrastrim) to patients with Fanconi anemia: a pilot study. *Blood* **88**:15888-93
- Scagni P et al (Ramenghi U) (**1998**) Use of recombinant granulocyte colony-stimulating factor in Fanconi´s anemia. *Haematologica* **83**:432-7
- Kumar M, Alter BP (**1998**) Hematopoietic growth factors for the treatment of aplastic anemia. *Curr Opin Hematol* **5**:226-34

Konventionelle Therapie: Knochenmarktransplantation mit Geschwister- und Verwandtenspendern

- Friedrich W, Ebell W et al (Kleihauer E) (**1987**) Therapie der Fanconi-Anämie durch Knochenmarktransplantation. *Monatsschr Kinderh* **135**:253
- Flowers ME et al (Thomas ED) (**1992**) Marrow transplantation for Fanconi anemia with or without leukemic transformation: an update of the Seattle experience. *Bone Marrow Transplant* **9**:167-73
- Kohli-Kumar M et al (Harris RE) (**1994**) Bone marrow transplantation in Fanconi anemia using matched sibling donors. *Blood* **84**:2050-4
- Gluckman E et al (**1995**) Bone marrow transplantation in Fanconi anemia. *Blood* **86**:2856-62
- Flowers ME et al (Storb R) (**1996**) Marrow transplantation for Fanconi anaemia: conditioning with reduced doses of cyclophosphamide without radiation. *Br J Haematol* **92**:699-706
- Solh H et al (Clink H) (**1997**) Bone marrow transplantation in patients with Fanconi anemia: experience with cyclophosphamide and total body irradiation conditioning regimen. *Pediatr Hematol Oncol* **14**:67-72
- Kapelushnik J et al (Nagler A) (**1997**) A fludarabine-based protocol for bone marrow transplantation in Fanconi´s anemia. *Bone Marrow Transplant* **20**:1109-10
- Guardiola P et al (Gluckman E) (**1998**) Allogeneic stem cell transplantation for Fanconi Anaemia. *Bone Marrow Transplant* **21**: Suppl 2:S24-7
- Socie G et al (Gluckman E) (**1998**) Transplantation for Fanconi´s anaemia: long-term follow-up of fifty patients transplanted from a sibling donor after low-dose cyclophosphamide and thoraco-abdominal irradiation for conditioning. *Br J Haematolog* **103**:249-55
- McCloy M et al (Dokal I) (**2001**) Fludarabine-based stem cell transplantation protocol for Fanconi´s anaemia in myelodysplastic transformation. *Br J Haematol* **112**:427-9
- Dufour C et al (**2001**) Stem cell transplantation from HLA-matched related donor for Fanconi´s anaemia: a retrospective review of the multicentric Italian experience on behalf of the AIEOP-GITMO. *Br J Haematol* **112**:796-805

- Ayas M et al (**2001**) Bone marrow transplantation from matched siblings in patients with Fanconi anemia utilizing low-dose cyclophosphamide, thoracoabdominal radiation and antithymocyte globulin. *Bone Marrow Transplant* **27**:139-43
- Maschan AA et al (**2004**) Fludarabine, low-dose busulfan and antithymocyte globulin as conditioning for Fanconi anemia patients receiving bone marrow transplantation from HLA-compatible related donors. *Bone Marrow Transplant* **34**:305-7
- Dalle JH et al (**2004**) Successful pregnancies after bone marrow transplantation for Fanconi anemia. *Bone Marrow Transplant* **34**:1099-100

Konventionelle Therapie: Knochenmarktransplantation mit Fremdspendern

- Davies SM et al (Weisdorf DJ) (**1996**) Unrelated donor bone marrow transplantation for Fanconi anemia. *Bone Marrow Transplant* **17**:43-7
- Zwaan CM et al (Vossen JM) (**1998**) Unrelated donor bone marrow transplantation in Fanconi anemia: the Leiden experience. *Bone Marrow Transplant* **21**:447-53

Konventionelle Therapie: Knochenmarktransplantation bei FA-Patienten mit AML bzw. MDS

- Guardiola P et al (Gluckman E) (**2003**) Effective graft-versus-leukemia effect after allogeneic stem cell transplantation using reduced-intensity preparative regimens in Fanconi anaemia patients with myelodysplastic syndrome or acute myeloid leukemia. *Br J Haematol* **122**:806-9
- Ayas M et al (**2004**) Allogeneic stem cell transplantation in patients with Fanconi´s anemia and myelodysplasia or leukemia utilizing low-dose cyclophosphamide and total body irradiation. *Bone Marrow Transplant* **33**:15-7

Konventionelle Therapie: Transplantation mit CD34-positiven Blutstammzellen

- Boulad F et al (**2000**) Stem cell transplantation for the treatment of Fanconi anaemia using a fludarabine-based cytoreductive regimen and T-cell-depleted related HLA-mismatched peripheral blood stem cell grafts. *Br J Haematol* **111**:1153-7
- Croop JM et al (Williams DA) (**2001**) Mobilization and collection of peripheral blood CD34+ cells from patients with Fanconi anemia. *Blood* **98**:2917-21
- Boyer MW et al (Harris RE) (**2003**) Low risk of graft-versus-host disease with transplantation of CD34 selected peripheral blood progenitor cells from alternative donors for Fanconi anemia. *J Pediatr Hematol Oncol* **25**:890-5

Komplikationen nach Knochenmark- bzw. Stammzelltransplantation

- Yakoub-Agha I et al (Gluckman E) (**2000**) Severe oesophagitis after allogeneic bone marrow transplantation for Fanconi´s anemia. *Bone Marrow Transplant* **26**:215-8
- Yalman N et al (**2001**) The effect of bone marrow transplantation on systemic and oral health in Fanconi´s aplastic anemia. *J Clin Pediatr Dent* **25**:329-32
- Trigg ME, Bond R (**2003**) Preparative therapies for those with Fanconi´s anemia undergoing stem cell transplants and subsequent engraftment rates. *Hematology* **8**:397-402
- Guardiola P et al (Gluckman E) (**2004**) Acute graft-versus-host disease in patients with Fanconi anemia or acquired aplastic anemia undergoing bone marrow transplantation form HLA-identical sibling donors: risk factors and influence on outcome. *Blood* **103**:73-7
- Barker J, Wagner JE (**2004**) Infectious disease complications after unrelated donor hematopoietic stem cell transplantation in children: Impact of stem cell source. *Biol Blood Marrow Transplant* **10**:737-8

Konventionelle Therapie: Nabelschnurblut-Transplantation

- Gluckman E et al (**1989**) Hematopoietic reconstitution in a patient with Fanconi´s anemia by means of umbilical-cord blood from an HLA-matched sibling. *N Engl J Med* **321**:1174-8
- Auerbach AD (**1994**) Umbilical cord transplants for genetic disease: diagnostic and ethical issues in fetal studies. *Blood Cells* **20**:303-9
- Aker M et al (Nagler A) (**1999**) Fludarabine-based protocol for human umbilical cord blood transplantation in children with Fanconi anemia. *J Pediatr Hematol Oncol* **21**:237-9
- Yoshimasu T et al (**2001**) Prompt and durable hematopoietic reconstitution by unrelated cord blood transplantation in a child with Fanconi anemia. *Bone Marrow Transplant* **27**:767-9
- Barker JN, Wagner JE (**2003**) Umbilical cord blood transplantation: current practice and future innovations. *Crit Rev Oncol Hematol* **48**:35-43
- Grewal SS et al (Wagner JE) (**2004**) Successful hematopoietic stem cell transplantation for Fanconi anemia from an unaffected HLA-genotype-identical sibling selected using preimplantation genetic diagnosis. *Blood* **103**:1147-51

Tumoren nach Knochenmarktransplantation

- Deeg HJ et al (Storb R) (**1996**) Malignancies after bone marrow transplantation for aplastic anemia and fanconi anemia: a joint Seattle and Paris analysis of results in 700 patients. *Blood* **87**:386-92

- Millen FJ et al (Swirsky D) (**1997**) Oral squamous cell carcinoma after allogeneic bone marrow transplantation for Fanconi anaemia. *Br J Haematol* **99**:410-4
- Jansisyanont P, Pazoki A, Ord RA (**2000**) Squamous cell carcinoma of the tongue after bone marrow transplantation in a patient with Fanconi´s anemia. *J Oral Maxillofac Surg* **58**:1454-7
- Rosenberg PS, Socie G, Alter BP, Gluckman E (**2005**) Risk of head and neck squamous cell cancer and death in patients with Fanconi anemia who did and did not receive transplants. *Blood* **105**:67-73

Auffälligkeiten bei Heterozygoten

- Auerbach AD, Wolman SR (**1978**) Carcinogen-induced chromosome breakage in Fanconi´s anaemia heterozygous cells. *Nature* **271**:69-71
- Swift M, Caldwell RJ, Chase C (**1980**) Reassessment of cancer predisposition of Fanconi anemia heterozygotes. *J Natl Cancer Inst* **65**:863-7
- Wunder E, Fleischer-Reischmann B (**1983**) Response of lymphocytes from Fanconi´s anemia patients and their heterozygous relatives to 8-methoxy-psoralene in a cloning survival test system. *Hum Genet* **64**:167-72
- Petridou M, Barret AJ (**1990**) Physical and laboratory characteristics of heterozygote carriers of the Fanconi aplasia gene. *Acta Paediatr Scand* **79**:1069-74
- Heim RA, Lench NJ, Swift M (**1992**) Heterozygous manifestations in four autosomal recessive human cancer-prone syndromes. *Mutat Res* **284**:25-36
- Mohan S et al (2000) Body proportions in Fanconi anemia heterozygotes. *Indian J Pediatr* **67**:797-801
- Rischewski JR et al (**2000**) A heterozygous frameshift mutation in the Fanconi anemia C gene in familial T-ALL and secondary malignancy. *Klin Padiatr* **212**:174-6
- Barquinero JF et al (**2001**) Cytogenetic sensitivity of three Fanconi anemia heterozygotes to bleomycin and ionizing radiation. *Cancer Genet Cytogenet* **124**:80-3

Untersuchungen der FA-Gene bei menschlichen Leukämien und Tumorerkrankungen

- Awan A et al (Eden T) (**1998**) Increased frequency of Fanconi anemia group C genetic variants in children with sporadic acute myeloid leukemia. *Blood* **91**:4813-4
- Cleton-Jansen AM et al (Cornelisse CJ) (**1999**) Mutation analysis of the Fanconi anemia A gene in breast tumors with loss of heterozygosity at 16q24.3. *Br J Cancer* **79**:1049-52
- Xie Y et al (Joenje H) (**2000**) Aberrant Fanconi anemia protein profiles in acute myeloid leukaemia cells. *Br J Haematol* **111**:1057-64
- Condie A et al (**2002**) Analysis of the Fanconi anaemia complementation group A gene in acute myeloid leukemia. *Leuk Lymphoma* **43**:1849-53

- van der Heijden MS et al (**2003**) Fanconi anemia gene mutations in young-onset pancreatic cancer. *Cancer Res* **63**:2585-8
- Olopade OI, Wei M (**2003**) FANCF methylation contributes to chemoselectivity in ovarian cancer. *Cancer Cell* **3**:417-20
- D'Andrea AD (**2003**) The Fanconi anemia/BRCA signaling pathway: disruption in cisplatin-sensitive ovarian cancers. *Cell Cycle* **2**:290-2
- Lensch MW et al (Bagby GC Jr) (**2003**) Acquired FANCA dysfunction and cytogenetic instability in adult acute myelogenous leukemia. *Blood* **102**:7-16
- Erdmann J (**2003**) Fanconi anemia research opens new doors in understanding of cancer. *J Natl Cancer Inst* **95**:1190-2
- Tischkowitz M et al (Joenje H) (**2003**) Bi-allelic silencing of the Fanconi anaemia gene FANCF in acute myeloid leukemia. *Br J Haematol* **123**:469-71
- Marsit CJ et al (**2004**) Inactivation of the Fanconi anemia/BRCA pathway in lung and oral cancers: implications for treatment and survival. *Oncogene* **23**:1000-4
- Rogers CD et al (**2004**) The genetics of FANCC and FANCG in familial pancreatic cancer. *Cancer Biol Ther* **3**:167-9
- Tischkowitz MD et al (Mathew CG) (**2004**) Deletion and reduced expression of the Fanconi anemia FANCA gene in sporadic acute myeloid leukemia. *Leukemia* **18**:420-5
- van der Heijden MS, Brody JR, Kern SE (**2004**) Functional screen of the Fanconi anemia pathway in cancer cells by fancd2 immunoblot. *Cancer Biol Ther* **3**:534-7
- Narayan G et al (**2004**) Promoter hypermethylation of FANCF: disruption of Fanconi anemia-BRCA pathway in cervical cancer. *Cancer Res* **64**:2994-7
- van der Heijden MS et al (**2004**) Functional defects in the Fanconi anemia pathway in pancreatic cancer cells. *Am J Pathol* **165**:651-7