

Index

Symbols

9p13 14, 20, 56
16q24.3 14, 28
176-307del 108, 109
176-307del clones 108
176-510del 108, 109
176-510del clones 108
322delG 8, 16, 18, 39, 40, 45, 46, 48, 49
796A/G 107
808C>T 110
868-869delFQ 106
890C/T 109
890C/T in exon 7 109
1059-1076del 109, 110
1066C>T 108-110
1066C>T mutation 109, 110
1235C/T 105, 107
1360-2014del 104, 106
1501A/G 107
1927C/G 106, 107
(1927C/G) in exon 22 106
2426G/A 107
2546delC 104, 105, 107, 112
3114C/G 107
3114C/T 107

A

α spectrin II 78
A-T 1-3, 5, 9, 10, 16
Acute myeloid leukemia (AML) 3, 4, 7-9, 13, 28, 32, 33, 40, 93
Acute nonlymphocytic leukemia 13
Adeno-associated virus (AAV) 96, 97
Adenosine deaminase 95
Adenoviruses 96
AKT 14
Akt kinase 77, 79
Alpha spectrin 16, 42
Alpha-tocopherol 83
Alu repeat sequences 29
AML/MDS 8
Androgens 4, 92, 94
Anti-FANCA antibody 69

Antioxidant 47, 48, 82-85
Antithymocyte globulin (ATG) 36, 93-96, 104
Aplastic anemia 1, 3-7, 13, 92, 93, 95, 96
Apoptosis 17, 32, 41-49, 75, 76, 79, 82, 97
Arabidopsis 61
Ashkenazi Jewish 10, 16, 29, 40
ASK signaling 18
ASK1 17, 18, 48, 49
 α SPII Σ^* 42, 78
Ataxia telangiectasia 1, 10, 62, 63
ATLD 5
ATM 2, 9, 10, 55, 63, 64
ATM kinase 63
ATR checkpoint kinase 19, 87
Autosomal 1, 36, 54, 67, 103
Autoubiquitination 70
Azathioprine 96

B

BARD1 62
BASC complex 63, 64
BLM 2, 21, 22, 55, 64, 70
Bloom syndrome (BS) 1, 2, 6, 70
Bone marrow (BM) 1, 3, 4, 7-9, 13, 15, 17, 20, 29, 30, 32, 37, 41, 43-48, 67, 75, 82, 84, 86, 87, 92-94, 96-98
Bone marrow failure 1, 3, 4, 7, 9, 13, 15, 29, 30, 32, 46, 75, 82, 84, 86, 87, 92, 97
Bone marrow transplant (BMT) 4, 30, 93-95, 98
Brazilian patient 105
BRCA1 2, 14, 16, 19, 21, 47, 58, 61-64, 68, 69, 75, 76, 78, 79, 112
BRCA2 2, 4, 7-10, 13, 14, 16, 18, 19, 21, 22, 55-58, 61-64, 68-70, 74, 79, 87, 103, 111
Breast cancer 9, 62-64, 75
BRG1 15, 16, 75, 76
BTB/POZ 75
Burst forming units-erythrocytes (BFU-E) 45, 93

C

c. 1059-1076del 110
C. elegans 61
 CAAT box 37
Café au lait 5, 9
 Carcinomas 1, 3, 4, 6, 8, 33, 95
 Caspase 41, 45-47, 76
 Caspase-3 activation 45-47
 Catalase 83-85
 CD4 17
 CD33 98
 CD34⁺ 37, 41, 42, 44, 45, 93-95, 97
 CD34⁺ *Fancc*^{-/-} 44
 CD95 44, 45
 Cdc2 16, 42, 43, 75
 Cdc2 kinase 42, 43
 cDNA 6, 14, 28, 36, 37, 39, 40, 43, 44, 46, 56, 61, 67, 104, 106-112
 cDNA library 37
 Central nervous system (CNS) 5, 8, 40, 54
 Chromatin 2, 15, 16, 18, 21, 22, 42, 75, 78
 Chromosome abnormalities 1, 2, 9
 Chromosome 7 7
 Chromosome 14 7
 Cis-platin 43, 82, 83
 CK-2 39
 Clinical trial 97
 Clonogenic assays 97
 CNS defects 40
 CNS malformations 8
 Coding sequence include G139E 40
 Colony forming units-granulocyte-macrophage (CFU-GM) 93
 Combined immunodeficiency disease 98
 Complementation analysis 6, 7, 14, 39, 54, 69
 CpG methylation-induced mutation 29
 Cr(VI) 84
 Cu/Zn superoxide dismutase 47
 Cyclooxygenase 2 84
 Cyclophosphamide (CP/CY) 82, 83, 93-96
 Cyclosporine 93, 95
 CYP2E1 57, 74

D

D195V 40
 Dehydroascorbic acid (DHA) 47
 DHPLC analysis 14
 Diepoxybutane (DEB) 2, 5, 6, 14, 36, 37, 44, 67, 74, 82, 83, 85, 103
 DNA 1, 2, 5-7, 9, 14, 18, 19, 21, 28, 31, 32, 36, 37, 42, 43, 46, 48, 55-58, 61-64, 67, 70, 71, 74-76, 78, 82-87, 93, 96, 97, 103, 104, 106, 108, 109, 111
 DNA crosslinking agent 1, 2, 5-7, 9, 36, 46, 67, 83, 93, 97, 103
 DNA damaging agents 57, 70, 96, 97
 DNA double strand break (DSB) 1, 2, 18, 46
 DNA excision repair 1
 DNA interstrand crosslinks 2, 46, 70, 111
 DNA repair 2, 18, 19, 21, 31, 32, 36, 37, 55, 58, 64, 76, 78, 82, 84, 86, 87, 96, 103
 DNA repair/cell cycle checkpoint 55
 DNA-dependent ATPase 75
Drosophila 61
 dsRNA 17, 36, 46

E

E3 ubiquitin ligase activity 62
 E417L 40
 Electron paramagnetic resonance (EPR) 84
 Endochondral ossification 40
 Endocrine abnormalities 82, 85
 Epidermal growth factor (EGF) 76
 ERCC1 21, 78, 112
 Ethnic background 30
 European Blood and Marrow Transplantation (EBMT) Group 93, 94
 European Fanconi Anemia Registry (EUFAR) 93
 Exon 3 15, 108
 Exon 3 (176-307del) 108
 Exon 4 16, 32, 39, 40, 108
 Exon 7 108, 109
 Exon 8 15, 43, 109, 110
 Exon 8 (1059-1076del) 109
 Exon 15
 Exon 22 106

F

FA knockout mice 15
 FA patients in Asian countries 103
 FA-A patient 7, 8, 28-31, 54, 107-109
 FA-C 7, 8, 17, 30, 36, 37, 39, 40, 42-49, 54, 67, 74, 75, 85, 103, 111
 FA-C lymphoblastoid cell line (FA-C LCL) 36, 37, 40, 42, 43, 45-48
 FA-G 7, 8, 17, 30, 54, 56, 57, 69, 109, 111, 112
 FA20P 104, 105
 FA33P 104
 FAAP95 2
 FANCA 2, 6, 8, 10, 13-17, 19-21, 28-33, 42, 54-57, 61-64, 67-71, 74-78, 82, 84, 97, 103-108, 111, 112
 FANCA exon 43 10
 FANCA kinase 14
 FANCA locus 105, 106
 Fanca murine models 32
 FANCA null mutation 30
 FANCA phosphorylation 31, 32, 55, 77
 FANCA-BRG1-BRCA1 75
 FANCB 2, 13-15, 20-22, 31, 55, 56, 58, 71, 111
 FANCC 2, 6-8, 10, 13-21, 29-31, 33, 36-49, 54-57, 61-63, 67-70, 71, 74, 75, 82, 84, 85, 97, 98, 103, 110, 111
 FANCC cDNA 40, 43, 44, 46, 111
 FANCC exon 14 7
Fancc^{-/-} mice 44-48
Fancc^{+/-} 44, 45
Fancc^{+/+} 44, 46
 FANCC322delG mutation 8
 FANCD1 1, 2, 4, 7, 9, 10, 13, 14, 16, 18, 21, 55-58, 61, 63, 64, 68, 69, 103, 111
 FANCD2 1, 2, 6, 7, 9, 10, 13-16, 18-22, 28, 33, 36, 41, 42, 46, 55-58, 61-64, 67-71, 75, 76, 78, 87, 103, 111
 FANCD2 ubiquitination 14, 20, 64
 FANCE 2, 6, 13, 14, 16, 19, 21, 31, 42, 48, 55-57, 62, 63, 67-70, 103, 111
 FANCF 2, 6, 13, 14, 19, 20, 31, 42, 55-57, 62, 63, 67-69, 71, 98, 103, 111
 FANCFORF 57
 FANCG 2, 3, 6, 13, 14, 16-21, 31, 33, 42, 55-57, 61-64, 67-71, 74, 75, 82, 84, 85, 103, 108-112

FANCG/CYP2E1 57
 FANCG/XRCC9 56
 FANCI 2, 14, 20, 55, 103
 FANCIJ 2, 7, 9, 14, 20, 55, 103
 FANCL 2, 6, 13-15, 20, 22, 28, 42, 55, 61-63, 69-71, 103
 FANCL/PHF9 70, 71
 FANCM 2, 103
 Fanconi anemia (FA) 1-10, 13-22, 28-33, 36-40, 42-49, 52-58, 61-65, 67-71, 74-76, 78, 79, 82-88, 92-98, 103-105, 107-109, 111, 112
 Fanconi anemia zinc finger (FAZF) 16, 42, 74, 75
 Fas ligand 17
 Fas ligation 36, 47, 48
 Fas receptor (CD95) 44, 45
 Fas-mediated cell death 45
 Fas-priming by IFN 47
 Flavin mononucleotide (FMN) 47
 Fludarabine 95, 96, 98
 Frameshift 7, 15, 28, 39, 40, 104, 105, 110
 Frameshift mutation 7, 28, 40, 104, 105
 Free radical 43, 47, 55, 57, 86
 Fungal infections 95

G

g. 1066C>T 110
 G1/S 41
 G2 phase of the cell cycle 36
 G2/M 41-43, 75, 97
 Gamma-irradiation 32, 41, 47
 Gene therapy 96-98
 Genotype-phenotype data 30, 32
 Glucose intolerance 86
 Glutathione (GSH) 47, 74, 83, 84, 86
 Glutathione S-transferase 47, 74, 83, 84
 Glutathione S-transferase (GSTP1) P1-1 16-18, 47-49, 74
 Graft versus host disease (GVHD) 93-96
 Granulocyte-colony stimulating factor (G-CSF) 92, 93, 97
 Grd19p 76
 GRP94 15, 16, 41, 75

H

3-hybrid 56, 69
 8-hydroxy deoxy guanosine (8-OHdG) 57, 84, 85
 70kDa heat shock protein (HSP70) 16, 17, 46, 48, 49, 84
 Haplotype analysis 18, 104
 Heat shock responses 20
 Hematological correction 96
 Hematopoiesis 3, 15-18, 22, 28-30, 32, 36, 40-45, 47, 48, 55, 75, 86, 92-94, 96-98
 Hematopoietic cells 16-18, 22, 41, 45, 47, 94, 96-98
 Hematopoietic stem cell (HPC) 29, 30, 36, 41, 43-45, 48, 49, 92-94, 96-98
 Hematopoietic growth factor 92, 93
 Heme oxygenase 83
 Hepatic tumors 92
 Hexavalent chromium compounds 82
 HIV-1 97
 HIV-2 97
 HLA-matched sibling 92, 93, 96
HMRE11 2, 5
 Homologous recombination (HR) 2, 19, 21, 32, 63, 68, 70, 71, 79, 112
 Homologous recombination repair 2, 19, 21, 68, 79
 Homozygous null (*Fancc*^{-/-}) mice 43
 HSC536 36, 39, 45, 46
 HSC536 cell line 39, 46
 HSC536 FA-C cell line 39
 Human immunodeficiency virus (HIV) 96, 97
 Hydrogen peroxide (H₂O₂) 18, 41, 47, 48, 57, 85
 Hydroxyl radicals 83
 Hygromycin 20
 Hyperglycaemia 85
 Hyperinsulinaemia 86
 Hypogonadism 17
 Hypomorphs 63
 Hypothyroidism 86
 Hypoxic 47

I

I312V 40
 ICL 41, 42, 46, 48, 111, 112
 IFN receptor- α 45
 I κ B kinase 15, 76
 IKK 15, 76-79
 IKK complex 76, 77, 79
 IKK signalsome 76, 78
 IKK2 15, 75, 76
 IL-3 45, 98
 Immunodeficiency 5, 6, 96, 98
 Immunosuppressive agent 95, 96
 Insertional mutagenesis 98
 Interferon gamma (IFN- γ) 17, 41, 44-49, 75, 85, 97
 International Bone Marrow Transplant Registry (IBMTR) 93
 International Fanconi Anemia Registry (IFAR) 3, 111
 Intragenic deletions 14
 Intron 4 7, 39, 40
 Intronless 19
 Ionizing radiation (IR) 1, 2, 6, 7, 9, 10, 19, 47, 62, 63, 70, 75
 IVS3+1G>C 20, 108, 109, 111, 112
 IVS4 + 4 A>T 39, 40
 IVS8-2A>G 110
 IVS13-1G>C 109
 IVS13-2A>G 108, 109
 IVS27-1G>A 104, 106
 IVS27-2A>T 104, 106
 IVS41-2A>G 104, 106

J

Jak kinase 17
 Japanese FA 10, 103, 104, 107, 109, 111, 112
 JNK 47, 48

L

L190F 40
 LCL 36, 37, 40, 46
 Lentivirus 96, 97
 Lentivirus HIV-1 97
 Lentivirus HIV-2 97
 Leukemia 1, 3, 4, 9, 10, 13, 32, 33, 54, 62, 75, 85, 86, 95, 96, 98

Lipid peroxidation 83, 84
 Lipoxygenase 86
LMO2 gene locus 98
 Lymphoblasts 37, 45, 46, 54, 56, 57, 67, 84
 Lymphopaenia 5
 Lymphoproliferative malignancies 96

M

8-methoxypsoralen 82, 84
 Macrophage inflammatory protein-1 α
 (MIP-1 α) 17, 44, 97
 MAPKKK 48
 Mass spectroscopy 62
 Medulloblastoma 4, 8, 9
 MEFs 45, 46, 48
 Melatonin 84
 Microcell fusions 61
 Microdeletions 14, 29
 Microinsertions 14
 Microphthalmia 32
 Mismatch repair 64
 Missense mutation 7, 9, 39, 104, 105
 Mitomycin C (MMC) 2, 5, 6, 8, 14, 18-20,
 22, 32, 36, 37, 39-47, 49, 54-57, 62, 63,
 67, 69, 70, 74, 82, 83, 85, 93, 97, 98,
 103, 111
 Mitosis 21, 41, 42, 75
 Modulatory role 68
 Monoubiquitination 2, 6, 7, 9, 13, 15, 21, 28,
 36, 41, 42, 46, 57, 61-64, 87
 Mosaicism 7, 18, 29, 97
 Mouse cells 9
 MRE11 21, 46, 55, 68, 112
 Multigenic disorder 13
 Murine leukemia virus (MLV) 96
 Mutation 1-3, 5-10, 13-22, 28-30, 32, 33,
 39-42, 45, 46, 49, 54-56, 61-64, 67-71,
 93, 103-112
 Mutation Q13X 39
 Mutation W22X 39
 Mvplp 76
 Myelodysplasia 13, 93
 Myelodysplastic syndrome (MDS) 3, 4, 7, 8,
 40
 Myelogenous leukemia 32, 85

N

N-acetylcysteine 83, 84
 NADPH 16, 47, 74, 84
 NADPH cytochrome P450 reductase 74, 84
 Nbs1 protein 5, 7
 Nephroblastoma 4
 Neutrophil 92, 93
 NF- κ B 57, 76, 77, 79
 Nijmegen breakage syndrome (NBS) 1, 2,
 5-7, 21, 46, 47, 55, 62, 68, 112
 Nitric oxide (NO) 47, 48
 Nitrogen mustard 43
 Nonerythroid α spectrin 112
 NonFanconi aplastic anemia 96
 Nonpathogenic polymorphic variants 107
 NSP2 57
 Nuclear translocation 16, 21
 Null mutations 2, 18, 30, 54, 68

O

Oligodeoxynucleotide 43
 Oxidative stress 14, 17, 18, 20, 47, 74, 82-87
 Oxygen 47, 74, 82, 83, 85
 Oxymethalone 4, 92

P

26S proteasome 41
 P211R 40
 p38 signalling cascades 48
 P450 47, 57, 74, 84, 86
 P450 2E1 57, 74, 84
 p53 37, 39, 43-45, 48, 75, 87
 Pancytopenia 3, 54, 86
 Patient AP78M 106
 Patient FA20P 104
 PHD finger protein-9 (PHF9) 14, 20, 69, 70,
 71
 PHD-type E3 ubiquitin ligase 70
 Phosphatidylinositol-3 kinase 31, 32
 Phosphatidylinositol-3,4,5-trisphosphate 77
 Phosphoinositide (PI) 76, 77
 Phosphoinositide 3-kinase (PI-3K) 77
 Phosphoinositide-dependent kinase-1 (PDK-1)
 77
 Phosphoprotein 39, 56
 Phosphorylation 9, 14, 15, 31, 32, 39, 46, 47,
 55, 63, 70, 75, 77-79
 PKC phosphorylation 39

PKR 16-18, 20, 46, 48, 49
see also RNA-dependent protein kinase
 Placental blood program 94
 Plasmid DNA 46
 Polymorphic site 104-107, 109
 Polymorphism 28-30, 38, 40, 55, 107, 109, 111
 Post-translational modification 14, 28, 70
 Preintegration complex 96
 Pro-oxidant state 82-86
 Promyelocytic leukemia zinc finger (PLZF) 42, 75
 Prostaglandin H synthase 86
 Protein ROM 19, 57
 Psoralen + UVA 42, 43
 PUVA 82, 84
 PX 76
 Pyrimidine dimers 1

Q

Q465R 40

R

RAD50 46, 55, 68, 112
 Rad51 16, 18, 19, 21, 22, 47, 56, 58, 62, 63, 69, 70
 Rad54 21
 Radiation therapy 93, 96, 97
 Reactive oxygen species (ROS) 47, 74, 76, 83, 84, 86
 RecQ helicase 70
 RED 47, 49, 74
 Redox cycling 47, 74, 82
 Redox regulation 18, 76
 Redox state 36, 47, 49, 84, 87
 Redox-related toxicity 82
 Restriction site of Cac8I 107
 Retroviral 44, 45, 96-98
 Retroviral vector 96-98
 Retrovirus 6, 44, 97
 Rev3 21
 RFLP analysis 104, 107, 108
 RMN assembly 46
 RNA splicing mutation 105
 RNA-dependent protein kinase 20, 46
see also PKR
 ROM 19, 57
 RPA 14/32/70 64
 RPA1 19
 Rutin 83

S

S-phase 2, 9, 16, 19, 21, 41-43, 46, 48, 62, 70
 S-phase checkpoint 9, 46
 S26F 40
 SCID 98
 SCID mouse xenotransplant 98
 Seckel syndrome 5
 Semiquinone radicals 83
 Serine 222 9, 63
 Serine kinase 14, 71
 Serine phosphorylation of FANCA 31
 Serine-threonine kinase 48
 Signal-regulating kinase 1 48
 Skin pigmentation 3-6, 82, 85, 86
 Snm1 21
 SNX1 76
 SNX5 16, 75-78
 Sod1 47
 Somatic mosaicism 29, 97
 Splicing mutation 14, 105, 109, 110
 Squamous cell carcinoma 4, 95
 STAT 17, 18, 46
 STAT molecules 17
 STAT1 phosphorylation 46, 75
 STAT1 signaling 45
 Stem cell transplantation 62, 92, 93
 Stoichiometry of FA proteins 31
 Superoxide dismutase (SOD) 47, 83
 SWI/SNF 15, 75, 76, 78, 79

T

T cell leukaemia 4, 9, 10
 T-cell 17, 94, 95, 98
 TATA box 37
 Testosterone 92
 TGF- β 41
 Thalidomide 86
 Thioredoxin (Trx) 83, 85, 86
 Thoracoabdominal 93, 94
 Thrombocytopenia 95
 Thrombocytosis 5
 Tip60 21
 TNF receptor 45
 Transgene 96-98
 Tumor necrosis factor alpha (TNF α) 17, 44-46, 49, 57, 76, 77, 85, 97
 Tyk2 17
 Type II diabetes mellitus 82

U

Ubiquitin ligase 2, 15, 20, 28, 61, 62, 70
Ultraviolet (UV) light 1, 82

V

V449M 40
V60I 40
Viral vectors 96, 97
Vitamin C 83
Vps5p 76

W

Wilms tumour 4, 9

X

X-linked FA 15
Xeroderma pigmentosum (XP) 1, 2
Xp22.31 14, 15, 58
XPF 21, 70, 78
XRCC2 21
XRCC3 21
XRCC9 2, 20, 56

Y

Yeast 2-hybrid 56, 57, 69
Yeast 3-hybrid 56