USMLE STEP 1

Pathology High Yield Associations

1. 11:22: transloc seen in ewing sarcoma
2. 14:18: follicular lymphoma IG heavy chain on 14, bcl-2 oncogene on 18 p95
3. 15:17: acute promyelocytic leukemia PML gene on 15 and RARa on 17 p95
4. 8:14: Burkitts c::myc is on 8, IG heavy chain is on 14 p95
5. 9:22: Philadelphia chromosome, CML bcr abl fusion c-abl on 9, bcr on 22 p95
6. Addison disease: primary adrenal failure p312
7. Albers SCHonberg dx: aka osteopetrosis, marble bone dx, dense skeleton, failure of osteoclastic activity p349
8. Alport syndrome: hereditary nephritis associated with nerve deafness and ocular disorders, mutation in gene for the a5 chain of type IV collagen p261
9. ANAs in SLE: dsDNA antibodies and Sm Smith antigen = highly specific for SLE p77
10. Angelman syndrome: materal transmission del(15)(q11q13), aka happy puppet syndrome, mental retardation, ataxia, seizures, inappropriate laughter p53
11. APC: tumor suppressor common in familial polyposis coli and adenoCA of colon as well as other tumors p96
12. Arnold Chiari formation: downward displacement of cerebellar tonsils and medulla through foramen magnum p364
13. Arthus reaction: seen in hyperacute rejection, = acute inflammation, fibrinoid necrosis of small vessels, and extensive thrombosis p72
14. Aschoff body: area of focal interstitial myocardial inflammation characterized by fragmented collagen and fibrinoid material, by large cells (Anitschkow myocytes) and occasional multinucleated giant cells (Aschoff cells) p140
15. Bax: gene product facilitates apoptosis p7
16. Bcl2: gene product inhibits apoptosis p7
17. Beckwith Weidemann syndrome: hemihypertrophy, macroglossia, organomegaly, neonatal hypoglycemia, various embryonal tumors, deleted WT 2 gene p268
18. Bense Jones protein: seen in urine of patients with multiple myeloma, Ig light chain either k or l p175
19. Berger dx: aka IgA nephropathy, deposition of IgA in mesangium, benign recurrent hematuria in kids following infection p261
20. Beriberi: this comes in a wet variety and a dry one, they are both Vit B1 thiamine deficiency
21. Bernard Soulier dx: autosomal recessive, unusually large platelets, lack of "GPIb IX V" p191
22. Binswanger dx: aka subcortical leukoencephalopathy, assoc with HTN, multiple lacunar infarcts and progressive demyelination in subcortical area 372
23. Birbeck granule: tennis racket shaped cytoplasmic inclusion seen in langerhans cells (so also seen in langerhan cell histiocytosis) p68
24. Bouchard nodes: osteophytes at PIP joints p355
25. Bowen dx: single erythematous plaque on shaft of penis or scrotum p280
26. Bowenoid papulosis: appears as multiple wart-like lesions resembling condyloma accuminatum p280
27. BRCA1: tumor suppressor mutated in breast and ovary CA p96
28. BRCA2: associated with breast alone (isn't one of these associated with prostate? Check) p96

29. Brenner tumor: ovarian tumor resembling bladder transitional epith p296
30. Brown tumor: non::neoplastic tumor like masses, fibrous replacement of resorbed bone p317
31. Brushfield spots: seen in Down synd, small white spots on periphery of iris p51
32. Brutons Agammaglobulinemia: x linked, absence of plasma cells and serum immunoglobulins, cell mediated immunity still ok, no germinal centers, recurrent bacterial infections, resistances to fungal and viral still, btk gene defect p73
33. Btk gene: defective in x-linked brutons agammaglobulinemia, B-cell tyrosine kinase p73
34. Budd Chiari: thrombotic occlusion of major hepatic veins, abnormal pain, jaundice, hepatomegaly, acites, liver failure, assoc with polycythemia vera, hepatocellular CA, and other common neoplasms, may occur as a complication of pregnancy p246
35. Buerger dx: aka thromboangiitis obliterans, acute inflame of small and med arteries of extremities, extending to adjacent veins and nerves, young jewish men, painful ischemic dx, worse with smoking p129
36. Burkitt lymphoma: aggressive B-cell lymphoma, EBV, starry sky appearance, c myc p179
37. C ANCA: associated with Wegener granulomatosis p128
38. Call Exner bodies: small follicles filled with eosinophilic secretion, diagnostic feature of granulose cell tumor p297
39. Charcot Bouchard aneurysm: aneurysm at small artery bifurcations p365
40. Charcot triad: nystagmus, intention tremor, scanning speech, seen in MS p370
41. Chediak Higashi syndrome: autosomal recessive, neutropenia, albinism, cranial and peripheral neuropathy, tendency to develop repeated infections, abnormal WBCs (abnormal mictotubul formation and large cytoplasmic granules = lysosomes) p25
42. Chromosome 10: paracentric inversion or translocation with 17 associated with papillary thyroid CA p316
43. Churg Strauss syndrome: aka allergic granulomatous angiitis, necrotizing vasculitis, variant of polyarteritis nodosa, involves pulmonary vasculature, peripheral eosinophilia, and asthma p128
44. Clue cell: vaginal epithelial cell with stippled appearance due to adherent coccobacilli p290
45. Codman triangle: radiologic appearance of periostium in bone tumors p352
46. Conn syndrome: primary aldosteronism p320
47. Cori dx: deficient debranching enzyme amylo-1,6-glucosidase, glycogen in liver, heart, skeletal muscle, stunted growth, hepatomegaly, hypoglycemia p57
48. Councilman body: small round eosinophilic masses found in dieing hepatocytes often containing chromatin remnants seen in viral hepatitis p7 and p242
49. Cri du chat syndrome: 5p chromosome deletion, severe mental retardation, microcephaly, catlike cry, low birth weight, round face, hypertelorism (wide eyes) p51
50. Crigler Najjar syndrome: severe unconj hyperbilirubinemia due to deficient glucuronyl transferase p239
51. Denys Drash syndrome: abnormal WT 1 gene, intersexual disorders, nephropathy, wilms tumor p266

52. DiGeorge syndrome: aka velocardiofacial syndrome and CATCH 22 syndrome, micro deletion of 22q11, Cardiac abnormalities, Abnormal facies, T cell deficit due to thymic hypoplasia, Cleft palate, Hypocalcemia due to hypoparathyroidism, from poor development of 3rd and 4th cranial arches p51 and p73
53. Donovan bodies: seen in granuloma inguinale, multiple organisms filling large histiocytes p291
54. Dry beriberi: peripheral neuropathy, atrophy of muscles, wet - high output cardiac failure, DCM, AV shunting, hypervolemia p116
55. Dubin Johnson syndrome: autosomal recessive, conj hyperbilirubinemia, defect in bili transport, black color to liver p239
56. E4 allele of apoprotein E: chromosome 19, allele common in alzheimers
57. Edwards syndrome: aka trisomy 18, mental retardation, prominent occiput, micrognathia (small jaw), low::set ears, rocker::bottom feet, finger deformities, congenital heart dx p51
58. Ewing sarcoma: small round blue cell tumor of bone, long bones, kids, 11:22 transloc p352
59. Fabry dx: aka angiokeratoma corporis diffusum universal, deficient a::galactosidase A, accumulate ceramide trihexoside, skin lesions angiokeratomas, fever, burning pain in extremities, cardiovascular and cerebrovascular involvement, death as adult by renal failure p59
60. Fanconi syndrome: renal tubular dysfunction, impaired reabsorption of glucose, amino acids, phosphate, and bicarb thus glycosuria, hyperphosphaturia, hypophosphatemia, aminoaciduria, systemic acidosis p264
61. Felty syndrome: variant of rheumatoid arthritis, splenmegally, neutropenia, RA p354
62. Ferruginous body: asbestosis, yellow-brown, rod shaped body with clubbed ends, stain with Prussian blue p204
63. FGFR3: gene mutated in achondroplasia (most common form of dwarfism), located at 4p16.3 p349
64. Foamy histiocyte: sphingomyelin containing phagocytes seen in Niemman::Pick dx in liver, spleen, lymph nodes and skin p57
65. Gardner syndrome: autosomal dominant, numerous adenomatous polyps along with osteomas and soft tissue tumors p230
66. Gaucher dx: deficient glucocerebrosidase, accumulation of glucocerebroside in cells of mononuclear phagocyte system 3 types :: see p 56
67. Gerstmann Straussler: Scheinker syndrome :: prion dx, fatal familial insomnia
68. Gilbert syndrome: modest elevated serum unconj bilirubin, due to decreased uptake by liver cells and reduced activity of glucuronyl transferase p239
69. GNAS1: mutation here causes pseudohypoparathyroidism p318
70. Goodpasture syndrome: aka antiglomerular basement membrane dx, antibodies against alveolar and glomerular basement membranes, linear immunoflorescence p261
71. Graves dx: autoimmune thyroiditis, hyperthyroid, exophthalmos, increased in HLA DR3 and HLA B8 pos people p315
72. Guillain Barre syndrome: acute inflammatory demyelinating dx primarily involving peripheral nerves p370
73. Hand Schuller Christian dx: aka chronic progressive histiocytosis, not as bad as Letterer Siwe p350
74. Hartnup dx: impaired tubular reabsorption of tryptophan, pellagra::like manifestations p264
75. Hashimoto thyroiditis: autoimmune hypothyroid, hurthle cells common p316

76. Heart failure cells: intra::alveolar hemosiderin-laden macrophages caused by RBCs from capillary rupture 2* to pulmonary HTN p35
77. Heberden nodes: osteophytes at DIP joints p355
78. Henoch Schonlein Purpura: hemorrhagic urticaria of extensor surgaces of arms, legs, and buttocks, with fever, arthralgias, and GI renal involvement similar to IgA nephropathy, associated with URIs p128 and 189
79. HER: 2/neu :: oncogene (aka c-erbB2), in breast cancer = poor prognosis p95
80. Hfe gene: located on xsome 6, mutation here = hereditary hemochromatosis p9
81. Hirano bodies: intracytoplasmic proximal dentritic eosinophilic inclusions consisting of actin seen in Alzheimers p370
82. Hirschsprung dx: aka congenital megacolon, dilation of colon due to absence of ganglion cells p227
83. HLA B27 antigen: associated with 90% cases of ankylosing spondylitis, common in rheumatoid arthritis p69 and p354
84. Hodgkin dx: malignant lymphoma with features resembling inflammatory disorder, young men, Reed Sternburg cells p176
85. Hunter syndrome: similar to Hurler, deficient L iduronosulfate sulfatase, accumulations of heparin sulfate and dermatan sulfate, hepatospenomegaly, micrognathia, retinal degeneration, joint stiffness, mental retardation, cardiac lesions p59
86. Huntington dx: autosomal dominant, fatal, progressive degeneration of striatum and frontal cortex with neuronal depletion and gliosis p372
87. Hurler syndrome: mucopolysaccharidosis, deficient a L iduronidase, accumulations of heparin sulfate and dermatan sulfate in heart, brain, liver, and other organs, progressive, hepatosplenomagaly, dwarfism, gargoyle-like facies, stubby fingers, corneal clouding, mental retardation, death by 10 years of age p57
88. Hurthle cell: epithelial cells with eosinophilc granular cytoplasm seen in hashimoto p316
89. Hutchinson freckle: aka lentigo maligna, precursor to lentigo maligna melanoma p336
90. If you've gotten this far, give yourself a high five: High 5
91. Kartagener syndrome: defective cilia, sinusitis, bronchiectasis, situs inversus, sometimes hearing loss and male sterility p202
92. Kawasaki dx: aka mucocutaneous lymph node syndrome, acute self-limited, infants and young kids, acute necrotizing vasculitis of small and medium vessels, fever hemorrhagic edema of conjunctivae, lips and oral mucosa, and cervical LAD, can cause coronary artery vasculitis with aneurysm formation p129
93. Kayser Fleischer ring: ring of copper deposited around iris seen in Wilson Dx p245
94. Kimmelsteil Wilson dx: diffuse nodular diabetic glomerulosclerosis p323
95. Kimmelstiel Wilson nodules: mesangial matrix material acculmulations seen in nodular glomerulosclerosis seen in diabetic nephropathy p259
96. Klinefelter syndrome: at least 2 X and one Y, hypogonadism, tall, gynecomastia, low testosterone, high pituitary gonadotropins, infertility, p51
97. Koilocyte: dysplastic cell seen in HPV p293
98. Krukenberg tumor: metastatic stomach CA to ovary p224

99. Lambert Eaton syndrome: paraneoplastic synd looks like myasthenia gravis, often small cell lung CA p347
100. Langhans giant cell: nuclei arranged in horseshoe-shaped pattern about periphery of cell, characteristic of but not specific for granulomatous TB (as opposed to a foreign body giant cell that has scattered nuclei) p26
101. Lesch Nyhan syndrome: deficient hypoxanthine::guanine phosphoribosyltransferase (HGPRT), low purine metabolism, high uric acid, thus gout, mental retardation, choreoathetosis, spasticity, self mutilation, aggressiveness p59
102. Letterer Siwe dx: aka acute disseminated langerhans cell histiocytosis, aggressive, usually fatal, kids hepatosplenomegaly, LAD, pancytopenia, pulmonary involvement, recurrent infections p350
103. Li Fraumeni syndrome: lots of tumors, breast CA, soft tissue sarc, brain tumors, leukemias, has loss of p53 tumor suppressor gene p96
104. Libman Sacks endocarditis: occurs in SLE, small vegitations on either or both surfaces of valves p141
105. Lisch nodule: pigmented iris hamartoma seen in neurofibromatosis
106. Lynch syndrome: hereditary nonpolyposis colon cancer, DNA repair genes messed up p96
107. Marfan syndrome: deficient fibrillin (constituent of microfibrils), arachnodactyly, ectopia lentis, aortic aneurysm, mitral valve prolapsed p55
108. McArdle syndrome: deficient muscle phosphorylase, glycogen in skeletal muscle, cramps with exertion p57
109. McCune Albright syndrome: polyostotic fibrous dysplasia, precocious puberty, café au lait spots, short stature, young girls p349
110. Mediterranean anemia or Cooley Anemia: B Thalassemia major p163
111. Meigs syndrome: triad of ovarian fibroma, ascites, hydrothorax p297
112. Munro abscesses: minute neutrophilic abscesses found within parakeratotic stratum corneum in psoriasis p333
113. N: myc :: neuroblastoma amplicatiion correlates inversely with degree of differentiation p95
114. Names of Vitamins: B1 thiamine, B2 riboflavin, B3 niacin, B6 pyridoxine, B12 cobalamin, C ascorbic acid, D calciferol, E a::tocopherol
115. Negri bodies: eosinophilic intracytoplasmic inclusions in hippocampus and purkinje cells seen in rabies infection p368
116. Nelson syndrome: development of large pituitary adenomas following bilat adrenalectomy p313
117. NF1: tumor suppressor mutated in Von Recklinhousen p96
118. Niemann Pick dx: deficient sphingomyelinase, accumulation of sphingomyelin in phagocytes, foamy histiocytes in liver, spleen, lymph nodes, skin, hepatosplenomegaly, anemia fever, occasional neuro degeneration, half have cherry red spot macula p 57
119. Orphan annie nuclei: seen in papillary thyroid CA p316
120. Osler Weber Rendu syndrome: aka hereditary hemorrhagic telangectasia, telangectasias in skin and mucous membranes, epistaxis, GI bleeds p55 and p189
121. p53: gene product facilitates apoptosis, decreases bcl::2 and increases Bax p7
122. p53: tumor suppressor gene, pretty much the coolest one p96
123. Paget dx of bone: aka osteitis deformans, increased osteoclastic and osteoblastic activity p348
124. Paget dx of breast: eczematoid lesion of nipple areola, large cells with clear “halo like” area, invade epidermis, underlying ductal CA p300

125. Paget dx of vulva: similar to paget breast, sometimes related to underlying adenoCA of sweat glands p291
126. Pancoast tumor: aka superior sulcus tumor, seen in bronchogenic CA, often with Horner's p 212
127. Patau syndrome: aka trisomy 13, mental retardation, microcephaly, micropthalmia, brain abnormalities, cleft lip/palate, polydactyly, rockerbottom feet, congenital heart dx p51
128. Peutz Jeghers syndrome: hamartomatous polyps in colon and small intest, freckles on lips, hands, genitalia, higher incidence of adeno CA of colon and other malignancies, stomach, breast, ovaries p230
129. Peyronie dx: subcutaneous fibrosis of dorsum of penis p279
130. PIG A: mutation = paroxysmal nocturnal hemoglobinuria p161
131. Plummer dx: combination of hyperthyroidism, nodular goiter, absence of exophthalmos p315
132. Plummer Vinson syndrome: iron deficient anemia associated with upper::esophageal web p157
133. Pompe dx: deficient a1,4 glucosidase, accumulation of glycogen in liver, heart, skeletal muscle, cardiomegaly, muscle hypotonia, spelnomegaly, intractable hypoglycemia, death from cardiorespiratory failure before age 3 p57
134. Potter sequence: oligohydramnios causes fetus to be smashed p257
135. Prader willi syndrome: paternal transmission del (15)(q11q13), hypogonad, hypotonia, mental retardation, behavior probs, uncrontrolled appetite p53
136. Ras oncogenes: oncogenes that code for p21 proteins which are membrane signalers p93
137. Raynaud disease: different in that it is the primary disorder, recurrent vasospasm of small arteries and venules, pallor, cyanosis, fingers and toes, young healthy women p129
138. Raynaud phenomenon: vasosmasm of small vessels, most often in fingers, seen with autoimmune dx like SLE, scleroderma and others (R in CREST), recurrent vasospasm, always secondary to underlying disorder p77 and p129
139. Rb gene: retinoblastoma on chromosome 13 p95 and p352
140. Reed Sternburg cell: binucleated or multinucleated giant cell seen in Hodgkin dx
141. Reidel thyroiditis: thyroid replacement by fibrous tissue, unkown origin, mimics CA p316
142. Reinke crystal: intracytoplasmic inclusions seen in leydig cell tumors p283
143. Reiter syndrome: urethritis, conjunctivitis, arthritis, associated with venereal or intestinal infection p354
144. ret: protooncogene mutated in medulary thyroid CA (MENII) p96
145. ret PTC: associated with papillary thyroid CA
146. Reye syndrome: aspirin toxicity seen in kids after acute febrile viral dx, microvesicular fatty change in liver and encephalopathy p108
147. Rotor syndrome: similar to Dubin Johnson with no black liver p239
148. Rouleaux formation: also seen in urine of MM, stacks of RBCs, will have high ESR p175
149. Sheehan syndrome: pituitary insufficiency post-partum, low TSH ACTH p298 and 312
150. Shy Drager syndrome: parkinsonism with autonomic dysfunction and orthostatic hypotension p373
151. Signet ring cell: mucinous cell, characteristic of stomach CA met to ovary (krukenberg tumor) p224

152. Simmonds dx: aka pituitary cachexia, generalized panhypopituitarism p312
153. Sipple syndrome: = MEN2a
154. Smudge cells: seen in CLL, leukemic B-cells that are fragile p172
155. Starry sky appearance: on lymph node biopsy, seen with Burkitt lymphoma p179
156. Stein Leventhal syndrome: aka polycystic ovary syndrome, young women, amenorrhea, infertility, obesity, hirsutism p295
157. Still disease: variant of rheumatoid arthritis, aka juvenile rheumatoid arthritis p354
158. Sturge Weber syndrome: port wine stain on face, ipsilateral glaucoma, vascular lesions of ocular choroidal tissue, extensive hemangiomatous involvement of meninges p337
159. Takayasu arteritis: aka pulseless dx, inflammation and stenosis of medium and large sized arteries, often aortic arch thus aortic arch syndrome p129
160. Tay Sachs dx: deficient hexosaminidase A, GM2 ganglioside accumulation, especially in neurons, CNS degeneration, mental/motor deterioration, blindness, cherry red spot on macula, death by 4 years of age p56
161. Turcot syndrome: adenomatous polyps with tumors of CNS p230
162. Turner syndrome: 45 XO, female hypogonadism, hypothyroid, short, webbed neck, 1* amenorrhea p52
163. Um… see p 94: there's like 15 different oncogenes listed, knock yourself out
164. Virchow node: superclavicular lymph node identifying metastatic stomach CA p224
165. Von economo encephalitis: infectious disorder, caused postencephalitic parkinsonism p373
166. Von Gierke dx: deficient glucose-6-phosphatase, accumulation of glycogen in liver and kidney, hepatomegaly, hypoglycemia p57
167. Von Hippel Lindau dx: hemangioblastoma or cavernous hemangioma of cerebellum, brainstem or retina, adenomas, cysts in liver, kidney, pancreas, and other organs, increased renal cell CA, gene = short arm of chromosome 3 p55 and p127
168. Von Recklinhousen dx: aka neurofibromatosis, neurofibromas in skin, schwannomas of CN VIII, café au lait spots, lisch nodules, skeletal disorders, other tumors, mutated NF1 tumor suppressor gene, osteolytic lesions, brown tumors p55 and p96 and 347
169. Waldenstrom Macroglobulinemia: manifestation of lymphplasmocytic lymphoma, B cell neoplasm p176
170. Waterhouse Friderichsen syndrome: catastrophic adrenal insuff and vascular collapse, hemorrhagic necrosis of adrenal cortex p321
171. Wegener granulomatosis: unknown etiology, necrotizing granulomatous vasculitis of small to medium sized vessels of the respiratory tract, kidneys, and other organs, circulating C-ANCAs p128
172. Werdnig Hoffman syndrome: aka infantile progressive spinal muscular atrophy, autosomal recessive, LMN disease, infants p373
173. Wermer syndrome: = MEN1
174. Wernicke Korsakoff syndrome: thiamine deficiency, cerebral dysfunction, aka alcoholic encephalopathy, hemorrhagic lesions in the mamillary bodies, confusion, ataxia, ophthalmoplegia, and memory loss plus confabulation p 105 and p115
175. Whipple dx: malabsorption syndrome, Tropheryma wippelii bacilli, small intestine commonly affected, arthralgias, cardiac, and neuro symptoms p227
176. Whipple triad: seen in insulinoma, episodic hyperinsulinemia and hypoglycemia, CNS dysfunction, reversal of CNS probs with administration of glucose p324
177. Wilms tumor: clear cell renal tumor seen in kids p266

178. Wilson dx: autosomal recessive, hepatitis, accumulation of copper due to transport problem, low ceruloplasmin seen, kayser fleischer rings in eyes p245

179. Wiskott Aldrich syndrome: x linked, aka immunodeficiency with thrombocytopenia and eczema, total immunoglobulins often normal, recurrent infections p74

180. WT1 and WT2: tumor suppressor mutated in wilms tumor p96

181. Zenker diverticulum: esophageal diverticulum just above upper esoph sphincter p221