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
Cri du chat syndrome	5p chromosome deletion, severe mental retardation, microcephaly, catlike cry, low birth weight, round face, hypertelorism (wide eyes) p51
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
Edwards syndrome	aka trisomy 18, mental retardation, prominent occiput, micrognathia (small jaw), low::set ears, rocker::bottom feet, finger deformities, congenital heart dx p51
Patau syndrome	aka trisomy 13, mental retardation, microcephaly, micropthalmia, brain abnormalities, cleft lip/palate, polydactyly, rockerbottom feet, congenital heart dx p51

Klinefelter syndrome	at least 2 X and one Y, hypogonadism, tall, gynecomastia, low testosterone, high pituitary gonadotropins, infertility, p51
Turner syndrome	45 XO, female hypogonadism, hypothyroid, short, webbed neck, 1* amenorrhea p52
Prader willi syndrome	paternal transmission del (15)(q11q13), hypogonad, hypotonia, mental retardation, behavior probs, uncrontrolled appetite p53
Angelman syndrome	materal transmission del(15)(q11q13), aka happy puppet syndrome, mental retardation, ataxia, seizures, inappropriate laughter p53
Osler Weber Rendu syndrome	aka hereditary hemorrhagic telangectasia, telangectasias in skin and mucous membranes, epistaxis, GI bleeds p55 and p189

Marfan syndrome	deficient fibrillin (constituent of microfibrils), arachnodactyly, ectopia lentis, aortic aneurysm, mitral valve prolapsed p55
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
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
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
Gaucher dx	deficient glucocerebrosidase, accumulation of glucocerebroside in cells of mononuclear phagocyte system 3 types :: see p 56

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
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
Von Gierke dx	deficient glucose-6-phosphatase, accumulation of glycogen in liver and kidney, hepatomegaly, hypoglycemia p57
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
Cori dx	deficient debranching enzyme amylo-1,6-glucosidase, glycogen in liver, heart, skeletal muscle, stunted growth, hepatomegaly, hypoglycemia p57

McArdle syndrome	deficient muscle phosphorylase, glycogen in skeletal muscle, cramps with exertion p57
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
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
Lesch Nyhan syndrome	deficient hypoxanthine::guanine phosphoribosyltransferase (HGPRT), low purine metabolism, high uric acid, thus gout, mental retardation, choreoathetosis, spasticity, self mutilation, aggressiveness p59
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

Wiskott Aldrich syndrome	x linked, aka immunodeficiency with thrombocytopenia and eczema, total immunoglobulins often normal, recurrent infections p74
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
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
Li Fraumeni syndrome	lots of tumors, breast CA, soft tissue sarc, brain tumors, leukemias, has loss of p53 tumor suppressor gene p96
Lynch syndrome	hereditary nonpolyposis colon cancer, DNA repair genes messed up p96

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
Reye syndrome	aspirin toxicity seen in kids after acute febrile viral dx, microvesicular fatty change in liver and encephalopathy p108
Beriberi	this comes in a wet variety and a dry one, they are both Vit B1 thiamine deficiency
Dry beriberi	peripheral neuropathy, atrophy of muscles, wet - high output cardiac failure, DCM, AV shunting, hypervolemia p116
Churg Strauss syndrome	aka allergic granulomatous angiitis, necrotizing vasculitis, variant of polyarteritis nodosa, involves pulmonary vasculature, peripheral eosinophilia, and asthma p128

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
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
Takayasu arteritis	aka pulseless dx, inflammation and stenosis of medium and large sized arteries, often aortic arch thus aortic arch syndrome p129
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
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

Libman Sacks endocarditis	occurs in SLE, small vegitations on either or both surfaces of valves p141
Plummer Vinson syndrome	iron deficient anemia associated with upper::esophageal web p157
Mediterranean anemia or Cooley Anemia	B Thalassemia major p163
Hodgkin dx	malignant lymphoma with features resembling inflammatory disorder, young men, Reed Sternburg cells p176
Waldenstrom Macroglobulinemia	manifestation of lymphplasmocytic lymphoma, B cell neoplasm p176

Burkitt lymphoma	aggressive B-cell lymphoma, EBV, starry sky appearance, c myc p179
Bernard Soulier dx	autosomal recessive, unusually large platelets, lack of “GPIb IX V” p191
Kartagener syndrome	defective cilia, sinusitis, bronchiectasis, situs inversus, sometimes hearing loss and male sterility p202
Pancoast tumor	aka superior sulcus tumor, seen in bronchogenic CA, often with Horner’s p 212
Zenker diverticulum	esophageal diverticulum just above upper esoph sphincter p221

Virchow node	superclavicular lymph node identifying metastatic stomach CA p224
Krukenberg tumor	metastatic stomach CA to ovary p224
Hirschsprung dx	aka congenital megacolon, dilation of colon due to absence of ganglion cells p227
Whipple dx	malabsorption syndrome, Tropheryma wippelii bacilli, small intestine commonly affected, arthralgias, cardiac, and neuro symptoms p227
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

Gardner syndrome	autosomal dominant, numerous adenomatous polyps along with osteomas and soft tissue tumors p230
Turcot syndrome	adenomatous polyps with tumors of CNS p230
Gilbert syndrome	modest elevated serum unconj bilirubin, due to decreased uptake by liver cells and reduced activity of glucuronyl transferase p239
Crigler Najjar syndrome	severe unconj hyperbilirubinemia due to deficient glucuronyl transferase p239
Dubin Johnson syndrome	autosomal recessive, conj hyperbilirubinemia, defect in bili transport, black color to liver p239

Rotor syndrome	similar to Dubin Johnson with no black liver p239
Wilson dx	autosomal recessive, hepatitis, accumulation of copper due to transport problem, low ceruloplasmin seen, kayser fleischer rings in eyes p245
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
Potter sequence	oligohydramnios causes fetus to be smashed p257
Goodpasture syndrome	aka antiglomerular basement membrane dx, antibodies against alveolar and glomerular basement membranes, linear immunoflorescence p261

Alport syndrome	hereditary nephritis associated with nerve deafness and ocular disorders, mutation in gene for the a5 chain of type IV collagen p261
Berger dx	aka IgA nephropathy, deposition of IgA in mesangium, benign recurrent hematuria in kids following infection p261
Fanconi syndrome	renal tubular dysfunction, impaired reabsorption of glucose, amino acids, phosphate, and bicarb thus glycosuria, hyperphosphaturia, hypophosphatemia, aminoaciduria, systemic acidosis p264
Hartnup dx	impaired tubular reabsorption of tryptophan, pellagra::like manifestations p264
Wilms tumor	clear cell renal tumor seen in kids p266

Denys Drash syndrome	abnormal WT 1 gene, intersexual disorders, nephropathy, wilms tumor p266
Beckwith Weidemann syndrome	hemihypertrophy, macroglossia, organomegaly, neonatal hypoglycemia, various embryonal tumors, deleted WT 2 gene p268
Peyronie dx	subcutaneous fibrosis of dorsum of penis p279
Bowenoid papulosis	appears as multiple wart-like lesions resembling condyloma accuminatum p280
Bowen dx	single erythematous plaque on shaft of penis or scrotum p280

Paget dx of vulva	similar to paget breast, sometimes related to underlying adenoCA of sweat glands p291
Stein Leventhal syndrome	aka polycystic ovary syndrome, young women, amenorrhea, infertility, obesity, hirsutism p295
Brenner tumor	ovarian tumor resembling bladder transitional epith p296
Meigs syndrome	triad of ovarian fibroma, ascites, hydrothorax p297
If you've gotten this far, give yourself a high five	High 5

Sheehan syndrome	pituitary insufficiency post-partum, low TSH ACTH p298 and 312
Paget dx of breast	eczematoid lesion of nipple areola, large cells with clear “halo like” area, invade epidermis, underlying ductal CA p300
Paget dx of bone	aka osteitis deformans, increased osteoclastic and osteoblastic activity p348
Simmonds dx	aka pituitary cachexia, generalized panhypopituitarism p312
Addison disease	primary adrenal failure p312

Nelson syndrome	development of large pituitary adenomas following bilat adrenalectomy p313
Graves dx	autoimmune thyroiditis, hyperthyroid, exophthalmos, increased in HLA DR3 and HLA B8 pos people p315
Plummer dx	combination of hyperthyroidism, nodular goiter, absence of exophthalmos p315
Hashimoto thyroiditis	autoimmune hypothyroid, hurthle cells common p316
Reidel thyroiditis	thyroid replacement by fibrous tissue, unkown origin, mimics CA p316

Conn syndrome	primary aldosteronism p320
Waterhouse Friderichsen syndrome	catastrophic adrenal insuff and vascular collapse, hemorrhagic necrosis of adrenal cortex p321
Kimmelsteil Wilson dx	diffuse nodular diabetic glomerulosclerosis p323
Whipple triad	seen in insulinoma, episodic hyperinsulinemia and hypoglycemia, CNS dysfunction, reversal of CNS probs with administration of glucose p324
Wermer syndrome	= MEN1

Sipple syndrome	= MEN2a
Sturge Weber syndrome	port wine stain on face, ipsilateral glaucoma, vascular lesions of ocular choroidal tissue, extensive hemangiomatous involvement of meninges p337
Lambert Eaton syndrome	paraneoplastic synd looks like myasthenia gravis, often small cell lung CA p347
McCune Albright syndrome	polyostotic fibrous dysplasia, precocious puberty, café au lait spots, short stature, young girls p349
Albers SCHonberg dx	aka osteopetrosis, marble bone dx, dense skeleton, failure of osteoclastic activity p349

Hand Schuller Christian dx	aka chronic progressive histiocytosis, not as bad as Letterer Siwe p350
Letterer Siwe dx	aka acute disseminated langerhans cell histiocytosis, aggressive, usually fatal, kids hepatosplenomegaly, LAD, pancytopenia, pulmonary involvement, recurrent infections p350
Ewing sarcoma	small round blue cell tumor of bone, long bones, kids, 11:22 transloc p352
Felty syndrome	variant of rheumatoid arthritis, splenmegally, neutropenia, RA p354
Still disease	variant of rheumatoid arthritis, aka juvenile rheumatoid arthritis p354

Reiter syndrome	urethritis, conjunctivitis, arthritis, associated with venereal or intestinal infection p354
Arnold Chiari formation	downward displacement of cerebellar tonsils and medulla through foramen magnum p364
Charcot Bouchard aneurysm	aneurysm at small artery bifurcations p365
Gerstmann Straussler	Scheinker syndrome :: prion dx, fatal familial insomnia
Charcot triad	nystagmus, intention tremor, scanning speech, seen in MS p370

Guillain Barre syndrome	acute inflammatory demyelinating dx primarily involving peripheral nerves p370
Binswanger dx	aka subcortical leukoencephalopathy, assoc with HTN, multiple lacunar infarcts and progressive demyelination in subcortical area 372
Huntington dx	autosomal dominant, fatal, progressive degeneration of striatum and frontal cortex with neuronal depletion and gliosis p372
Von economo encephalitis	infectious disorder, caused postencephalitic parkinsonism p373
Shy Drager syndrome	parkinsonism with autonomic dysfunction and orthostatic hypotension p373

Werdnig Hoffman syndrome	aka infantile progressive spinal muscular atrophy, autosomal recessive, LMN disease, infants p373
Councilman body	small round eosinophilic masses found in dieing hepatocytes often containing chromatin remnants seen in viral hepatitis p7 and p242
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
Heart failure cells	intra::alveolar hemosiderin-laden macrophages caused by RBCs from capillary rupture 2* to pulmonary HTN p35
Brushfield spots	seen in Down synd, small white spots on periphery of iris p51

Lisch nodule	pigmented iris hamartoma seen in neurofibromatosis
Foamy histiocyte	sphingomyelin containing phagocytes seen in Niemman::Pick dx in liver, spleen, lymph nodes and skin p57
Birbeck granule	tennis racket shaped cytoplasmic inclusion seen in langerhans cells (so also seen in langerhan cell histiocytosis) p68
Arthus reaction	seen in hyperacute rejection, = acute inflammation, fibrinoid necrosis of small vessels, and extensive thrombosis p72
Names of Vitamins	B1 thiamine, B2 riboflavin, B3 niacin, B6 pyridoxine, B12 cobalamin, C ascorbic acid, D calciferol, E a::tocopherol

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
Smudge cells	seen in CLL, leukemic B-cells that are fragile p172
Bense Jones protein	seen in urine of patients with multiple myeloma, Ig light chain either k or l p175
Rouleaux formation	also seen in urine of MM, stacks of RBCs, will have high ESR p175
Reed Sternburg cell	binucleated or multinucleated giant cell seen in Hodgkin dx

Starry sky appearance	on lymph node biopsy, seen with Burkitt lymphoma p179
Ferruginous body	asbestosis, yellow-brown, rod shaped body with clubbed ends, stain with Prussian blue p204
Signet ring cell	mucinous cell, characteristic of stomach CA met to ovary (krukenberg tumor) p224
Kayser Fleischer ring	ring of copper deposited around iris seen in Wilson Dx p245
Kimmelstiel Wilson nodules	mesangial matrix material acculmulations seen in nodular glomerulosclerosis seen in diabetic nephropathy p259

Reinke crystal	intracytoplasmic inclusions seen in leydig cell tumors p283
Clue cell	vaginal epithelial cell with stippled appearance due to adherent coccobacilli p290
Donovan bodies	seen in granuloma inguinale, multiple organisms filling large histiocytes p291
Koilocyte	dysplastic cell seen in HPV p293
Call Exner bodies	small follicles filled with eosinophilic secretion, diagnostic feature of granulose cell tumor p297

Hurthle cell	epithelial cells with eosinophilc granular cytoplasm seen in hashimoto p316
Orphan annie nuclei	seen in papillary thyroid CA p316
Brown tumor	non::neoplastic tumor like masses, fibrous replacement of resorbed bone p317
Munro abscesses	minute neutrophilic abscesses found within parakeratotic stratum corneum in psoriasis p333
Hutchinson freckle	aka lentigo maligna, precursor to lentigo maligna melanoma p336

Codman triangle	radiologic appearance of periostium in bone tumors p352
Heberden nodes	osteophytes at DIP joints p355
Bouchard nodes	osteophytes at PIP joints p355
Negri bodies	eosinophilic intracytoplasmic inclusions in hippocampus and purkinje cells seen in rabies infection p368
Hirano bodies	intracytoplasmic proximal dentritic eosinophilic inclusions consisting of actin seen in Alzheimers p370

Bcl2	gene product inhibits apoptosis p7
Bax	gene product facilitates apoptosis p7
p53	gene product facilitates apoptosis, decreases bcl::2 and increases Bax p7
Hfe gene	located on xsome 6, mutation here = hereditary hemochromatosis p9
HLA B27 antigen	associated with 90% cases of ankylosing spondylitis, common in rheumatoid arthritis p69 and p354

Btk gene	defective in x-linked brutons agammaglobulinemia, B-cell tyrosine kinase p73
ANAs in SLE	dsDNA antibodies and Sm Smith antigen = highly specific for SLE p77
Ras oncogenes	oncogenes that code for p21 proteins which are membrane signalers p93
Um… see p 94	there's like 15 different oncogenes listed, knock yourself out
8:14	Burkitts c::myc is on 8, IG heavy chain is on 14 p95

11:22	transloc seen in ewing sarcoma
14:18	follicular lymphoma IG heavy chain on 14, bcl-2 oncogene on 18 p95
9:22	Philadelphia chromosome, CML bcr abl fusion c-abl on 9, bcr on 22 p95
15:17	acute promyelocytic leukemia PML gene on 15 and RARa on 17 p95
HER	2/neu :: oncogene (aka c-erbB2), in breast cancer = poor prognosis p95

N	myc :: neuroblastoma amplicatiion correlates inversely with degree of differentiation p95
Rb gene	retinoblastoma on chromosome 13 p95 and p352
p53	tumor suppressor gene, pretty much the coolest one p96
NF1	tumor suppressor mutated in Von Recklinhousen p96
WT1 and WT2	tumor suppressor mutated in wilms tumor p96

APC	tumor suppressor common in familial polyposis coli and adenoCA of colon as well as other tumors p96
BRCA1	tumor suppressor mutated in breast and ovary CA p96
BRCA2	associated with breast alone (isn't one of these associated with prostate? Check) p96
ret	protooncogene mutated in medulary thyroid CA (MENII) p96
C ANCA	associated with Wegener granulomatosis p128

PIG A	mutation = paroxysmal nocturnal hemoglobinuria p161
Chromosome 10	paracentric inversion or translocation with 17 associated with papillary thyroid CA p316
ret PTC	associated with papillary thyroid CA
GNAS1	mutation here causes pseudohypoparathyroidism p318
FGFR3	gene mutated in achondroplasia (most common form of dwarfism), located at 4p16.3 p349

E4 allele of apoprotein E	chromosome 19, allele common in alzheimers