Appendix E
Diseases and References Used in Project-Based Cell Biology Class (2000–2001)[a,b]

Disease	Organelle	Protein, gene	Human interest story; symptoms	Review of organelle or process	Review of disease	Research papers
Adrenoleuko dystrophy	Peroxisome	ALDP, ABCD1 gene	http://www.mtsu.edu/jsanborn/dslwl.htm OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=300100	(Purdue and Lazarow, 2001) (Gottesman and Ambudkar, 2001)	(Moser, 2000) (Gould and Valle, 2000)	(Liu *et al.*, 1999) (Chang *et al.*, 1999) (Zolman *et al.*, 2001)
Alpha-1 antitrypsin deficiency	ER, secretion	Alpha-1 antitrypsin, AAT	http://www.alphaone.org/alpha1/special_stories/shirley_dennis.htm OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi? id=107400	(Janciauskiene, 2001) (Bross *et al.*, 1999) (Harter and Reinhard, 2000)	(Eriksson, 1999) (Coakley *et al.*, 2001) (Stockley, 2001)	(Burrows *et al.*, 2000)
Alport's disease	Extracellular matrix	Collagen IV, COL4A5	http://www.usatoday.com/life/health/doctor/lhdoc079.htm OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=301050		(Myllyharju and Kivirikko, 2001) (McCarthy and Maino, 2000)	(Harvey *et al.*, 2001) (Heikkila *et al.*, 2001) (Heidet *et al.*, 2000)
Angelman syndrome	Ubiquitination	ANCR, UBE3A	http://www.self-determination.org/newsletter1249/newsletter_show.htm-doc_id=10124.htm OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=105830	(Wilkinson, 2000) (Depraetere, 2001)	(Williams *et al.*, 2001) (Cassidy *et al.*, 2000)	(Nawaz *et al.*, 1999)
Duchenne muscular dystrophy	Plasma membrane	Dystrophin, DMD	http://www.mdausa.org/publications/Quest/q53mccoys.html OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=310200	(Jones *et al.*, 2000; McGowan and Marinkovich, 2000; Patton, 2000) http://www.neuro.wustl.edu/neuromuscular/musdist/dag2.htm	(Dubowitz, 2000) (Chamberlain and Benian, 2000) (Winder, 2001)	(Bartlett *et al.*, 2000)
Emery–Dreifuss muscular dystrophy	Nuclear matrix	Emerin, EMD	http://www.jhu.edu/jhumag/1100web/witch.html OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=181350	(Gruenbaum *et al.*, 2000)	(Hutchison *et al.*, 2001) (Nagano and Arahata, 2000) (Morris, 2000) (Wilson, 2000)	(Wilson, 2000)
Epidermolysis bullosa	Extracellular matrix	Keratin, KRT5, KRT14	http://www.debra.org/Patient%20Support/article1.htm OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=131900	(Parry and Steinert, 1999) (Herrmann and Aebi, 2000)	(Fuchs, 1998) (Fuchs and Cleveland, 1998)	(Peters *et al.*, 2001)

Disease	Organelle	Protein, gene	Human interest story; symptoms	Review of organelle or process	Review of disease	Research papers
Fibrodysplasia ossificans progressiva	Signaling	Not determined, possibly NOG, BMP4	http://www.eagletribune.com/news/stories/19990801/NH_003.htm or http://wire.ap.org/APpackages/fop/index.html OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=135100	(Miyazono *et al.*, 2001)	(Reddi, 1997) (Nakayama *et al.*, 2000) (Christian, 2000)	(Helvering *et al.*, 2000)
Machado–Joseph disease	Ubiquitin-like protein	Ataxin 3, ATX3	http://irmas.freeyellow.com/index.html http://ijdfnonprofit.freeyellow.com/newfile.html OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=109150	(Johnson, 2000) (Weissman, 2001) (Mayer, 2000)	(Yamada *et al.*, 2000). (Evidente *et al.*, 2000)	(Chan *et al.*, 2000; Warrick *et al.*, 1999) (Davidson *et al.*, 2000)
MERRF (myoclonus epilepsy with ragged red Æbers)	Mitochondria	Mitochondrial genes	http://www.umdf.org/personaljourneys/olivia_steele.html http://www.mdausa.org/publications/Quest/q64mito.html OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=545000	http://www.neuro.wustl.edu/neuromuscular/mitosyn.html	http://biochemgen.ucsd.edu/mmdc/eptoc.htm (Howell, 1999) (Thorburn and Dahl, 2001) (Orth and Schapira, 2001)	(Yasukawa *et al.*, 2001) (James *et al.*, 1999) (Enriquez *et al.*, 2000)
Tay–Sachs disease	Lysosomes	Hexos aminadase, HEXA	http://www.geocities.com/enchantedforest/pond/3061/colton.html OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=272800		(Mahuran, 1999) (Okada and O'Brien, 2001) (Suzuki, 2001)	(Adamali *et al.*, 1999) (Guidotti *et al.*, 1999)
Treacher Collins syndrome	Nucleolus	Treacle, TCOF1	http://www.treachercollins.org/main.html OMIM: http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=154500	(Schwarzacher and Mosgoeller, 2000)	(Marsh and Dixon, 2000)	(Splendore *et al.*, 2000) (Isaac *et al.*, 2000) (Dixon *et al.*, 2000)

[a] Abbreviations: ALDP = X-linked adrenolcukodystrophy protein; OMIM = Online Mendelian Inheritance in Man (web site); ANCR = Angelman Syndrome Chromosomal Region; ER = endoplasmic reticulum; NOG = noggin.

[b] References:

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