

## Appendix G

### Comparison Chart of Features of Human Diseases

<i>Genetics</i>	1: EDMD	2: MJD	3: TCS	4: ALD
Gene(s) name				
Chromosomal location				
Inheritance (autosomal? X-linked? dominant or recessive?)				
In what types of cells and when is gene expressed?				
Number & types of mutations				
Evolutionarily conserved? To what degree?				
<i>Protein</i>	1: EDMD	2: MJD	3: TCS	4: ALD
Size, number of amino acids?				
Location of protein in the cell				
Protein binds to or interacts with ...				
Structural features of protein				
Protein function				
In what classes of organisms is this protein found? Explain.				
<i>Organismal features of disease</i>	1: EDMD	2: MJD	3: TCS	4: ALD
What are the major symptoms of disease in LAY TERMS?				
Related genetic diseases?				
What types of cells are affected? Why?				
Unusual features of disease				
Animal models for disease				
<i>Cellular &amp; molecular biology of disease</i>	1: EDMD	2: MJD	3: TCS	4: ALD
What organelle or process is affected in this disease? What is the normal function of this organelle or process?				
In what types of cells, organisms, kingdoms, etc., is this organelle (process) found? What evolutionary variations exist?				
How do the mutations alter function of the organelle or process, resulting in disease symptoms? What is the connection between the mutation and the symptoms?				
<i>Relationships to your disease</i>	1: EDMD	2: MJD	3: TCS	4: ALD
Compare and contrast features of this disease to the one you are researching.				
<sup>a</sup> Abbreviations: EDMD = Emery–Dreifuss muscular dystrophy; MJD = Machado–Joseph disease; TCS = Treacher Collins Syndrome; ALD = adrenoleukodystrophy.				