Bioinformatics

Find the answers to the following questions:

PART ONE:

1. Open Genecards (http://www.genecards.org) database
2. Use keyword «aniridia»
3. What is the gene name that is causing disorder named Aniridia?
4. What is Aniridia?
5. What is genomic location of this gene (use „Jump to“ menu on the left)
6. How many mRNA (transcription variants) of these gene is predicted by RefSeq database?
7. How many transcripts are predicted by the Alternative Splicing Database (ASD)?
8. How many transcripts are predicted by Ensembl?
9. How would you rationalize any discrepancy?
10. How many protein isoforms of this gene is Uniprot database predicting?
11. The Ensembl Accession Number for the aniridia gene
12. How many human Pax genes is there?
13. Define «paralogs»?
14. How many orthologs is there in a mouse and how many in Drosophila?
15. Define «orthologs»?
16. What is the function of this gene?
17. Can you tell something about the tissues expression of this gene?
18. What is the product of this gene? In which cellular processes is involved?
19. Is aniridia monogenic or polygenic disorder?
20. What is OMIM database?
21. Find Aniridia in OMIM. What is the accession number for aniridia?
22. What types of mutations that resulted in aniridia are described (look under „Molecular genetics“)?
23. Is there any tumor often associated with aniridia?

PART II.

1. Open Uniprot database (http://www.uniprot.org).
2. Use HER2 keyword. Select human HER2.
3. What is the function of HER2 protein?
4. Where in the cell is this protein localized?
5. Does HER2 exists in the cell in a monomeric or dimeric form?
6. How many isoforms of HER2 is there?
7. To see the differences in isoforms, make «alignment»?
8. What are known interaction partners of HER2 protein?
9. Can you find the structure of HER2 protein in PDB database (there is a link in Uniprot database)?
10. Are there any drugs to be used for HER2 treatments?