CONCEPT MAP OF GENETIC DISORDERS (most of this info is also in your notes. Some information is added here so you can understand examples of each category. If you don't see a Genetic Disorders disease in your notes, you won't have to know it for the exam.) Two categories—chromosomal aberrations & single gene disorders Chromosomal aberrations Key characteristics (XC): Single gene disorders (next page) · one or more chromosomes damaged often caused by clastogens such as xravs, drugs, viruses, free radicals Alterations in structure KC: 2 sub-categories: alterations in number (aneuploidy) when parts of chromosomes are deleted, duplicated, or rearranged one type of rearrangement is called translocation alterations in structure an example of translocation-- Philadelphia chromosome. Alterations in number (aneuploidy) KC: Philadelphia chromosome -> CML caused by non-disjunction-failure to divide properly KC: non-disjunction results in too few chromosomes (45) or too results from translocation. many (47): part of a particular chromosome "breaks off" & attaches to another. · the left over, deformed, piece is known as the Philadelphia too fewtoo manychromosome monosomy polysomy If a person has this chromosome, will more likely have. CML—chronic myelocytic leukemia · CML occurs because the genes on the once-healthy Turner's Syndrome Trisomy 21 (Down's syndrome) chromosome control WBC production-now those genes KCC: RC: damaged -> WBC production "goes crazy" in bone marrow -> 31" chromosome set has 3. presence of only: huge number of WBCs in blood (leukocytosis)-> diagnosis of instead of two & this causes. 45 chromosomes leukemia. -one X instead of XX malfunction of the genes that code for normal development. 58/5 (phenotypel): mental retardation epicanthic fold. short limbs large tongue-

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