

<p>1</p> <ul style="list-style-type: none"> - Gaucher - L-myc gene <i>(involved in small cell carcinoma)</i> - Presenilin 2 (Alz) 	<p>2</p>	<p>3</p> <ul style="list-style-type: none"> - Von Hippel Lindau <i>(VHL)</i> 	<p>4</p> <ul style="list-style-type: none"> - Huntington <i>(CAG)</i> - Achondroplasia <i>(FGFR3 gene)</i> 	<p>5</p> <ul style="list-style-type: none"> - Cri-Du-Chat <i>(5p-)</i> - APC <i>(FAP)</i> 	<p>6</p> <ul style="list-style-type: none"> HFE gene Hemochromatosis
<p>7</p> <ul style="list-style-type: none"> - Cystic Fibrosis <i>(CFTR)</i> 	<p>8</p> <ul style="list-style-type: none"> - C-myc gene <i>(involved in Burkitt's lymphoma)</i> 	<p>9</p> <ul style="list-style-type: none"> - P 16 Tumor suppressor gene (Melanoma) - Friedreich's Ataxia <i>(Fratxin gene GAA triple repeats)</i> 	<p>10</p>	<p>11</p> <ul style="list-style-type: none"> - Nieman Pick <i>(sphingomyelinase)</i> - Beta Globin Chain <i>(β-Thal)</i> - BCL-1 Gene <i>(cyclin D)</i> 	<p>12</p>
<p>13</p> <ul style="list-style-type: none"> - Patau Trisomy - RB Gene <i>(retinoblastoma and osteosarcoma)</i> - BRCA 2 	<p>14</p> <ul style="list-style-type: none"> - Presenilin 1 <i>(alzheimer)</i> 	<p>15</p> <ul style="list-style-type: none"> - Tay-Sachs <i>(Hexosaminidase A)</i> - Marfan <i>(FBN1)</i> - Prader-Willi & Angelman <i>(imprinting)</i> 	<p>16</p> <ul style="list-style-type: none"> - PCKD1 <i>(polycystic kidney disease – polycystin1)</i> - α Globin Chain <i>(Alpha Thal)</i> - Tuberous Sclerosis (TSC2) 	<p>17</p> <ul style="list-style-type: none"> - NF1 <i>(Neurofibromatosis type 1 – Von Recklinghausen)</i> - P 53 -BRCA 1 	<p>18</p> <ul style="list-style-type: none"> - Edwards Trisomy - BCL 2 - DCC <i>(colon)</i>
<p>19</p> <ul style="list-style-type: none"> - Familial Hyper Cholesterolemia <i>(LDL Receptor mutation)</i> 	<p>20</p>	<p>21</p> <ul style="list-style-type: none"> - Down's Trisomy - B-APP Protein <i>(Alzheimer)</i> - SOD1 <i>(superoxide dismutase – altered in ALS)</i> 	<p>22</p> <ul style="list-style-type: none"> - NF2 <i>(Neurofibromatosis 2 – Merlin protein)</i> - DiGeorge Syn. <i>(locus 11 deletion)</i> 	<p>23</p>	<p>Baronerocks.com</p>

X	Quantitative Sexual Chromosome Diseases	Y
<ul style="list-style-type: none"> - Menkes Syndrome (ATP7A Copper Efflux Protein) - Fragile X Syndrome (CGG on gene FMR-1) - Bruton's (BTK gene) Agammaglobulinemia - Lesch-Nyhan (HPRT1) <ul style="list-style-type: none"> - Wiskott Aldrich (WASP Protein) -SCID (IL receptor mutation) - Duchenne MD (Dystrophin) <ul style="list-style-type: none"> - Alports (COL4A5) - Hemophilia A&B (F8&9) 	<ul style="list-style-type: none"> - Klinefelter Syndrome XXY (Barr Body) - Turner Syndrome XO (Shox Gene) 	<p>Baronerocks.com</p>

Translocations
<p>9:22 : Philadelphia (Bcr-Abl over expression. P210KD Tyrosine Kinase. In CML it provides better prognosis, in ALL it provides worst prognosis.)</p>
<p>8:14 : Burkitt's Lymphoma (C-myc gene over expression)</p>
<p>11:14 : Mantle Cell Lymphoma (Bcl-1 gene Aka Cyclin D)</p>
<p>14:18 : Follicular NHL (Bcl-2)</p>
<p>15:17 : AML M3 (Auer Rods, DIC)</p>
<p>11:22 : Ewing's Sarcoma</p>
<p>12:21 : Provides better prognosis in ALL</p>