

10 - 7. Mendelian inheritance

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© SPMM Course Disorder Location and mode of transmission Features DiGeorge (Velocardiofacial) 22q11.2 Autosomal dominant, 50% risk to offsprings, 5-10% risk of deletion in parents. If offspring has the deletion, then 25% chance of schizophrenia, if not then general population risk ~1%. Mild to moderate learning disability, facial deformities esp. cleft palate, absent or malformed parathyroids resulting in hypocalcemia, broad nasal bridge, articulatory speech and swallowing problems, >25% have psychosis Williams syndrome 7q11 microdeletion Hypercalcemia at birth, supra valvular aortic stenosis, moderate learning disability, disinhibited disposition, speech that appears superficially fluent, hyperacusis. Smith Magenis syndrome 17p11.2 microdeletion Moderate to severe learning disability, self harming behaviours e.g., pulling off nails (onychotillomania) and inserting foreign bodies into body orifices. Sleep disturbances and self hugging are also noted. Angelman syndrome Deletion of 15q11-13 maternally inherited (see genomic imprinting below) Developmental delay, low IQ, jerky movements especially hand-flapping, frequent smiling, and seizures. Prader-Willi syndrome Deletion of 15q11-13 paternally inherited (see genomic imprinting below) Obesity, short stature, small limbs, decreased IQ with hyperphagia and skin picking. Cri-du-chat syndrome Deletion of chromosome 5p (the locus 5p15.2 is responsible for the phenotype) Feeding problems due to difficulty swallowing and sucking, cat-like cry with poorly developed facial features.

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Johann Mendel was a Catholic priest who was interested in horticulture and botany. He studied garden peas and proposed 'laws' of inheritance. The first law is the law of uniformity. According to this law, if two plants that differ in just one trait (black and white) are crossed, then the resulting hybrids will be uniform in the chosen trait (either black or white, not blue). This is not entirely true as later geneticists demonstrated intermediate phenotypes resulting from co-dominant heterozygous expression. The second law is called the principle of segregation. It states that "for any particular trait, the pair of alleles of each parent separate and only one allele passes from each parent on to an offspring. Which allele in a parent's pair of alleles is inherited is a matter of pure chance". For example if there are two alleles one determining black colour and the other determining white in mother and two alleles with one

