

Concise Guide to Dysmorphology

Condition	Physical characteristics (head to toe)		Neurobehavioral effects/developmental challenges		Support Resources for Families
22q deletion syndrome	<ul style="list-style-type: none"> Doesn't look like their parents Small relative to family members Cleft palate or talks like they have a cleft palate even without structural problems 	<ul style="list-style-type: none"> Hypernasal speech Immunodeficiency Cardiac defects Congenital heart malformation 	<ul style="list-style-type: none"> Major psychiatric problems in 20% of cases at adolescence/young adulthood, eg schizophrenia, depression, anxiety 	<ul style="list-style-type: none"> Learning challenges High risk for social difficulties 	The 22q Family Foundation: https://22qfamily-foundation.org
Alcohol-related neurodevelopmental disorder (ARND)	Not likely to see specific facial features.	<p><i>For full fetal alcohol syndrome:</i></p> <ul style="list-style-type: none"> Small head circumference Small palpebral fissures Epicanthal folds Upturned nose Flat nasal bridge and midface area Smooth philtrum Thin upper lip 	<p><i>Neurocognitive challenges in:</i></p> <ul style="list-style-type: none"> Executive function Long term memory formation Working memory 	<p><i>Self-regulation challenges in:</i></p> <ul style="list-style-type: none"> Sensory processing Motor planning Emotional regulation <p><i>Adaptive functioning challenges in:</i></p> <ul style="list-style-type: none"> Flexible thinking for social problem solving 	Families Affected by Fetal Alcohol Spectrum Disorder: https://fafasd.net
Fragile X	<ul style="list-style-type: none"> Prominent brow Long, narrow face Large ears 	<ul style="list-style-type: none"> Prominent jaw Macroorchidism after puberty in males Flat feet 	<ul style="list-style-type: none"> Significant cognitive challenges in males, more subtle learning challenges in females 	<ul style="list-style-type: none"> Anxiety Higher prevalence in those with other at-risk family members 	The National Fragile X Foundation: https://fragilex.org/
Sex chromosome aneuploidy conditions	<ul style="list-style-type: none"> Usually no facial differences Possibly wide spaced eyes, epicanthal folds Clindactyly (curving) of fifth finger Hyperextensible joints Long limbs Flat feet 	<p><i>For extra x males: small testicles (eg Klinefelter Syndrome)</i></p> <p><i>For Turners' syndrome females:</i></p> <ul style="list-style-type: none"> Short stature Webbing of neck Broad chest Shortened fourth finger Cardiac defects 	<ul style="list-style-type: none"> Developmental delays Learning disabilities 	<ul style="list-style-type: none"> Emotional dysregulation Delayed puberty 	The Association for X and Y Variations (AXYS): https://genetic.org/about/
Trisomy 21	<ul style="list-style-type: none"> Almond-shaped eyes with white spots on the irises Epicanthal folds Small ears 	<ul style="list-style-type: none"> Flattened nose Protruding tongue Shortened neck Small hands and feet 	<ul style="list-style-type: none"> Intellectual disability Speech and language delays Obstructive sleep apnea Congenital heart defects Hypothyroidism Celiac disease 	<ul style="list-style-type: none"> Hearing loss Poor coordination Autism Attention-deficit/hyperactivity disorder Psychosis Early dementia persons 	National Down Syndrome Society: https://ndss.org/

Editor's note: Educational and support resources for other rare chromosome and gene disorders not covered here may be found at: <https://rarechromo.org/>