

ABOUT C22C



Chromosome 22 was the first chromosome to be sequenced in the Human Genome Project. Beyond the conditions listed in this brochure, chromosome 22 has also been researched as a link to bi-polar disorder, schizophrenia, and some forms of cancer.

Started in 1997 with just a handful of families, C22C has expanded to represent all families who face issues involving chromosome 22. We offer a quarterly newsletter, parent network registry, informative and interactive website, offer e-mail support lists, hold family gatherings, and promote research studies. Visit our website today and join us!

www.c22c.org



A few of our families at the July 2009 family reunion in Niagara Falls



Bringing Families TOGETHER!

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22Q11 DELETION

SYNDROME

People with 22q11 deletion syndrome have a tiny piece of chromosome 22 missing at this breakpoint (q11.2). This syndrome is also commonly referred to as Velo-cardio-facial Syndrome and DiGeorge Syndrome. Features of the syndrome may include heart anomalies, palatal anomalies (clefts, velopharyngeal insufficiency) learning difficulties, emotional/behavioural issues, hypocalcaemia, immune deficiency, and other concerns. 22q11 deletion is estimated to occur in 1 in 2000-4000 births.



22q11 MICRODUPLICATION—A new syndrome recognized in 2003, this condition is highly variable. Some features may include developmental and speech delays, congenital anomalies, and behaviour issues. Features may overlap with the 22q11 deletion syndrome.

22Q13 DELETION /PHELAN-MCDERMID SYNDROME

This deletion occurs at the very end tip (The distal end) of Chromosome 22. People missing this part of chromosome 22 may have hypotonia, developmental delays, speech difficulties, seizures, and ear abnormalities as well as other possible complications.

RING 22

Chromosome 22 Ring is a very rare disorder, which is caused by the breakage of one copy of chromosome 22 at each of its ends, and then reconnecting to form a ring. The amount of genetic material that is lost when the chromosome breaks can determine the severity of symptoms. The most common findings in this disorder are mental delay, speech problems, hypotonia (muscle weakness) and lack of coordination.

EMANUEL SYNDROME AND THE 11/22

TRANSLOCATION

This disorder has been referred to as partial trisomy 22, unbalanced 11/22 translocation, or supernumerary der (22) syndrome. It typically causes moderate to severe mental deficiency, and some children can be affected by heart defects, hypotonia, cleft palate, kidney, genital and ear abnormalities, as well as other complications.

The unbalanced translocation is usually inherited from a parent, who carries a balanced translocation between these two chromosomes. Translocation between the 11th and 22nd chromosomes is the most common reciprocal translocation in man. Carriers have a higher risk of experiencing miscarriages (approx. 30% rate) and a risk (up to 10%) of having a child affected by Emanuel Syndrome.



CAT EYE SYNDROME

(can be referred to as CES, partial tetrasomy 22, partial trisomy 22, inverted duplication (22qter-22q11) or Schmid-Fraccaro Syndrome)

CES is a very rare syndrome. It is named so because approximately 50% of people born with CES have a coloboma in the eyes, which makes their eyes appear like “cat’s eyes”. Some features may include anal atresia, unilateral or bilateral iris coloboma, ear pits or tags, cardiac defects, kidney abnormalities, short stature, skeletal problems, mild-moderate mental impair-

ment, cleft palate. More rare malformations can affect almost any organ.

MOSAIC TRISOMY 22

Mosaic Trisomy 22 is a rare chromosome 22 disorder in which the chromosome is present three times instead of two, only in SOME cells of the body. The severity of the disorder is dependent upon the percentage of cells affected. Features may include microcephaly (small head) abnormal ears, webbed neck, cardiac and kidney problems, growth retardation, mental delay, and hemidystrophy (abnormal development of each side of the body) as well as other complications.

COMPLETE TRISOMY 22

Complete or Full trisomy 22 occurs when an extra (third) copy of this chromosome is present in every cell of a person’s body, when normally there should only be two copies. Most affected babies with complete trisomy 22 pass away before birth or shortly after due to severe birth defects. Complete trisomy 22 is the second most common finding in miscarriages, after trisomy 16. Survival beyond the first trimester of gestation is very rare. The syndrome causes severe malformations.

ORPHAN 22 DISORDERS

There are other rare disorders which can involve chromosome 22. Some of these may involve translocations with other chromosomes, or deletions or trisomies of chromosome 22. Although literature is rare for many of these rare conditions, families can benefit from networking with other families. Our group has several “unique” families registered.