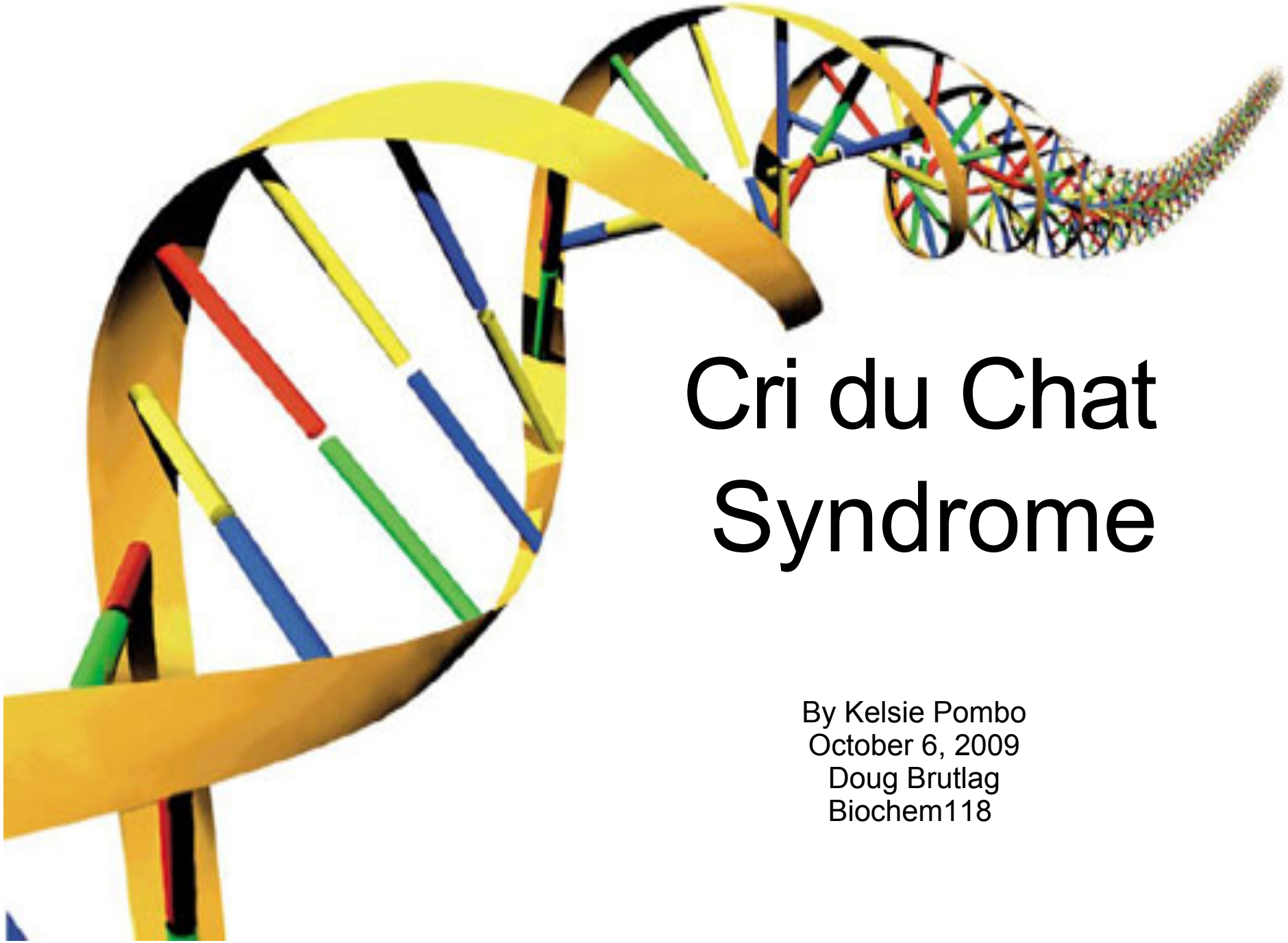
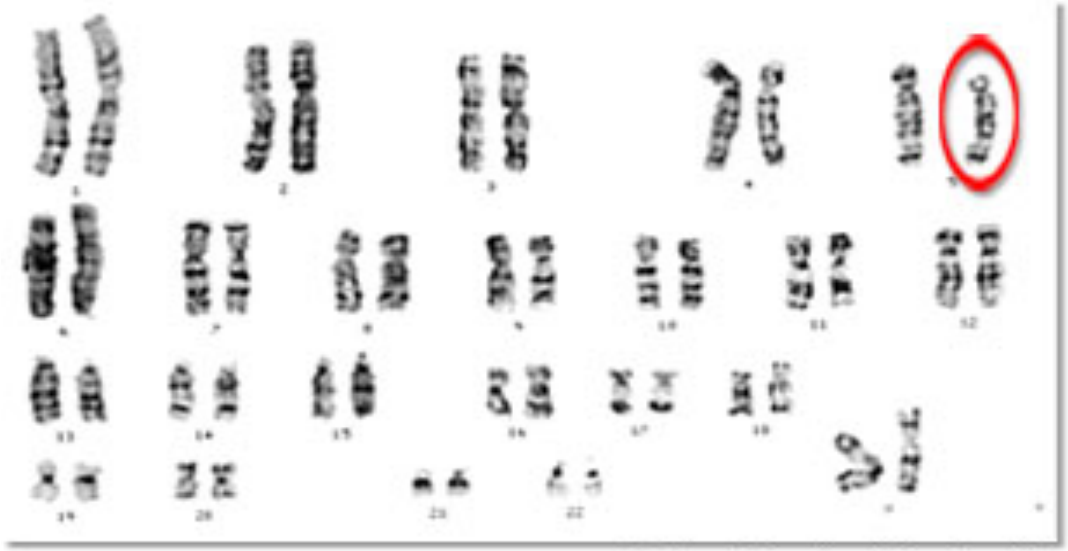


Cri du Chat Syndrome

By Kelsie Pombo
October 6, 2009
Doug Brutlag
Biochem118

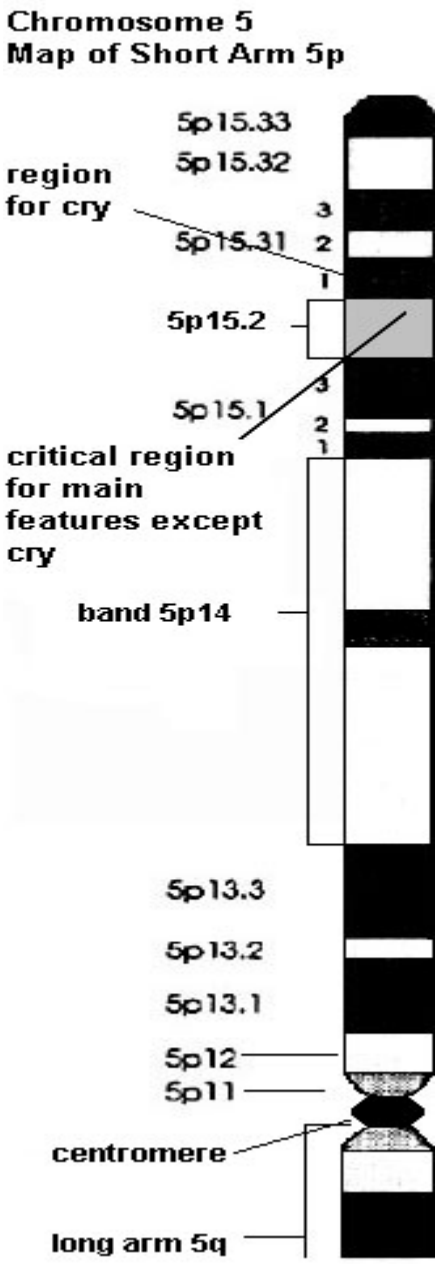
"5p-Syndrome"

- 1 in 20,000 to 50,000 newborns have syndrome.
- Caused by a deletion of the end of the short (p) arm of chromosome 5.
  - 90% caused by random deletions during meiosis.
  - 10-15% inherited from parents with balanced translocations.
  - Affected people typically have no history of disease in family.
  - 
    Tokyo Medical University
- Found in people of all ethnic backgrounds.
- Slightly more common in females.

The Genetics...

- **CTNND2** - “catenin (cadherin-associated protein), delta 2 (neural plakophilin-related arm-repeat protein).”
  - Located on 5p15.2
  - Provides instructions for making protein called **delta-catenin**.
  - Plays role in cell-cell adhesion, cell movement, neuronal migration, and brain development.
  - May be important in function of neural synapses.

- **TERT gene**
  - Localized to 5p15.33
  - Codes for a protein that is the rate-limiting component for telomerase activity.



Diagnosis

- High pitched “cry of the cat”
- Physical characteristics:
  - Microcephaly (Small head size)
  - Hypertelorism (widely-spaced eyes)
  - Broad, low nasal ridge
  - Low birth weight
  - Hypotonica (weak muscle tone)
  - High palate, round face, lowset ears
  - Folds of skin over upper eyelids
  - Distinctive creases in palms
  - Webbing on hands and feet
- Behavioural Characteristics:
  - Significant retardation
  - Feeding difficulties
  - Delays in walking; scoliosis
  - Hyperactivity

Symptoms:

- Rare serious heart defects, organ abnormalities, and other life-threatening medical conditions.
- Intellectual disability
- Difficulty with language (particularly expression)
- Visual and hearing impairments
- Delayed motor development
- Short attention span
- Hyperactivity
- Obsessive or repetitive behaviors

Treatment:

- Physiotherapy
- Hearing aids
- Surgical treatment for visual problems
- Speech pathology
- Educational intervention
- Behavioural management
- Ongoing support from family and friends
- (http://video.yahoo.com/watch/269765/1828426)

Future...

- Early intervention programs
  - Enhancement of social and sensory development
- Early diagnosis of mild cases
- Prenatal diagnosis
- Collaboration between developmental therapists and geneticists
- Gene therapy (in far future)

References:

Genetics Home Reference, Cri-du-chat syndrome.
http://ghr.nlm.nih.gov/condition=criduchatsyndrome

Genetic Home Reference, Genes: CTNN2
http://ghr.nlm.nih.gov/gene=ctnnd2

OMIM, Cri du Chat
http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=123450

Cri du Chat Support Group of Australia
http://www.criduchat.asn.au/criduchat/links.htm

Zhang, A. et al. Deletion of the telomerase reverse transcriptase gene and haploinsufficiency of telomere maintenance in cri du chat syndrome. *Am. J. Hum. Genet.* 72: 940-948, 2003.
http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?cmd=entry&id=123450#123450_Reference15