Disease Presentation:
DiGeorge Syndrome

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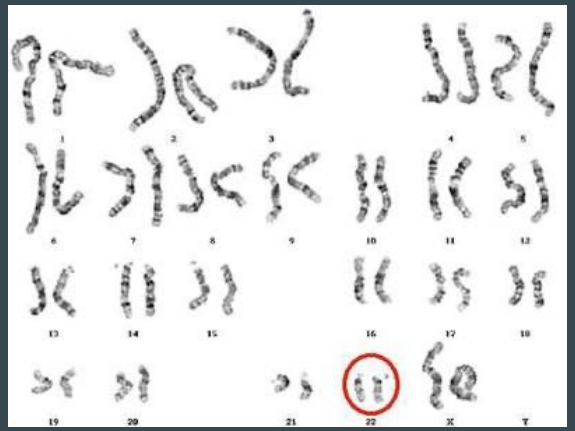


Discussion of Mutation

Heterozygous deletion of 22q11.2

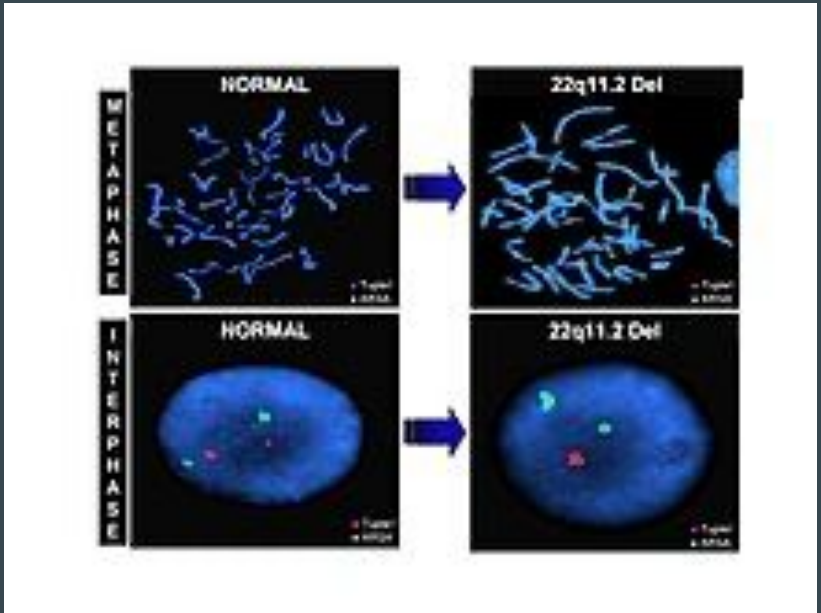
- Found on the 22 chromosome

Many importances found on this locus

- Embryonic development

Genes missing in this locus include

- TBX1 Gene
- COMT Gene



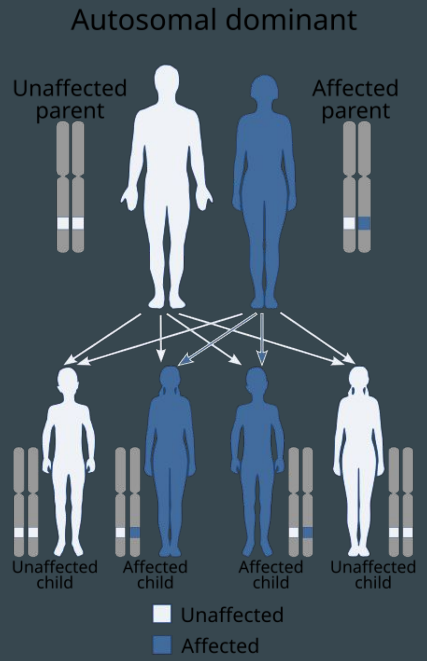
Discussion of Transmission

Mutation

- Most common and completely random
- At the time of fertilization or embryonic development

Inheritance

- Heterozygous deletion
- Passed from affected mother or affected father



Symptoms

Complete DGS

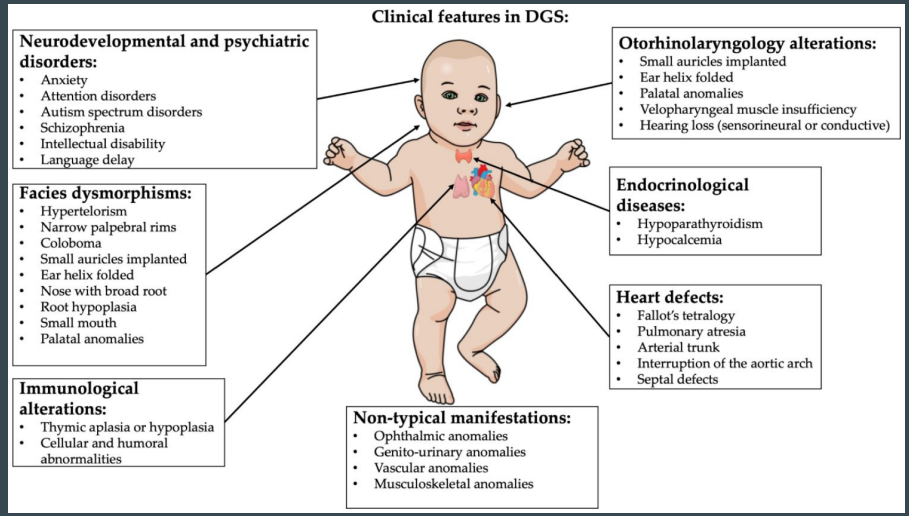
- Fatal during infancy

Partial DGS

- Diagnosed based on clinical phenotypes

Clinical phenotypes

- developmental delays
- Feeding difficulties
- Craniofacial feature deformities
- Psychiatric problems



Epidemiology

Most common deletion syndrome

- Affects 0.1% of fetuses
- 1 in 4,000-6,000 live births

More common than believed

- Patients undiagnosed
- Genetic testing is unavailable

More research is needed

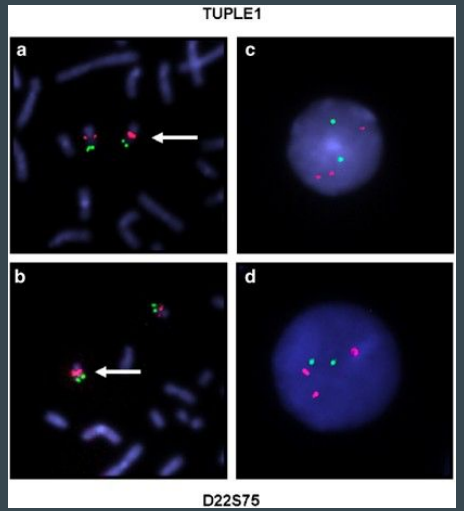
Screening

Many different ways to test

- Cytogenetic high resolution banding
- Restriction fragment length polymorphism

FISH

- Fluorescence in situ hybridization
- Rapid, simple, less invasive
- More effective in identifying this



Treatment

No current cure for this disorder

Deficits found in individuals

- Patients with this undergo testing
- This testing could help with treatment

Autoimmune defects associated with DGS

- Testing when given vaccines

Citation

Bajaj, S., Thombare, T. S., Tullu, M. S., & Agrawal, M. (2016). " FISHed" out the diagnosis: A case of DiGeorge syndrome. *Journal of Postgraduate Medicine*, *62*(2), 118-123.

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