Down syndrome: Bridging Genes, Brain and Cognition

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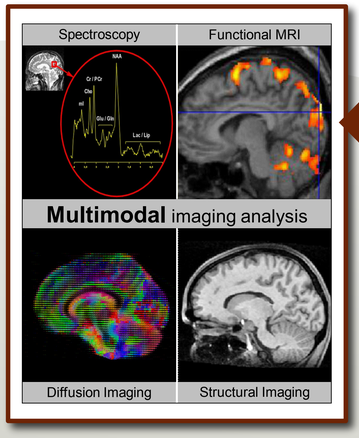
Down Syndrome
Occur in 1/800 births
> 300,000 in USA
> $2.1 B/year

Intellectual Disability ADHD
OCD Depression
Heart Disease Seizures
Alzheimer's Disease Leukemia

Treatment Targets Brain Circuits
Drug Development Diagnostics
Translate to Medicine Diagnostics & Treatment



Integration of Neuroscience and Genetics

Neuroimage Analysis

MRI/DTI MRS

Neural Imaging Twins

DS Learning and Memory

Patient Recruitment

NIH Grant
$3 Million, 2011-16
Down Syndrome: Bridging Genes, Brain, and Cognition

Biomarkers for Translational Treatments of Intellectual Disabilities

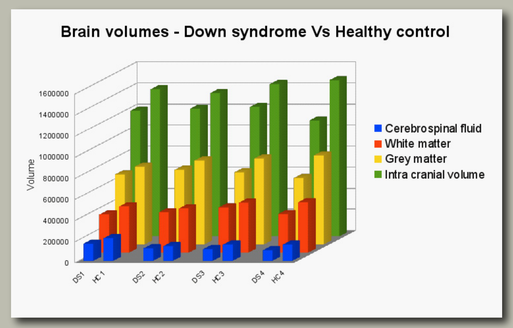
Generate Core Expertise

Utah Recognized Among Premier Centers for Neurodevelopmental Disorder Imaging & Treatment

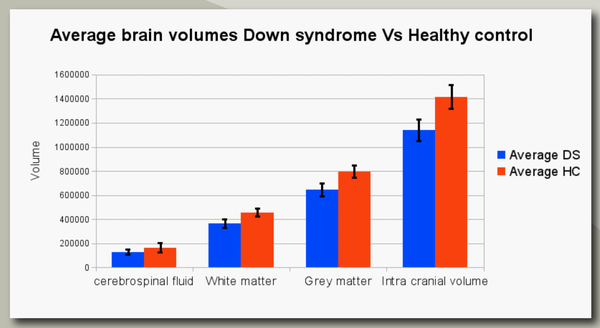
Brain volume analysis

Typical Down Syndrome phenotype includes different trajectory of neuroanatomy development. Analysis of these differences will give insight into genotype/phenotype/behavior relationships and influence of genetics on brain development.

Analysis performed on 8 Down syndromes (DS) and 9 healthy subjects (HC).
► Average brain volumes of the 2 population shows significant differences for all tissues.
► Age/sex paired subjects (DS vs HC) show consistent differences.



Volumes of brain tissues, fluid and intra-cranial space for four pairs of age and sex matched subjects: one Down syndrome and one healthy subject



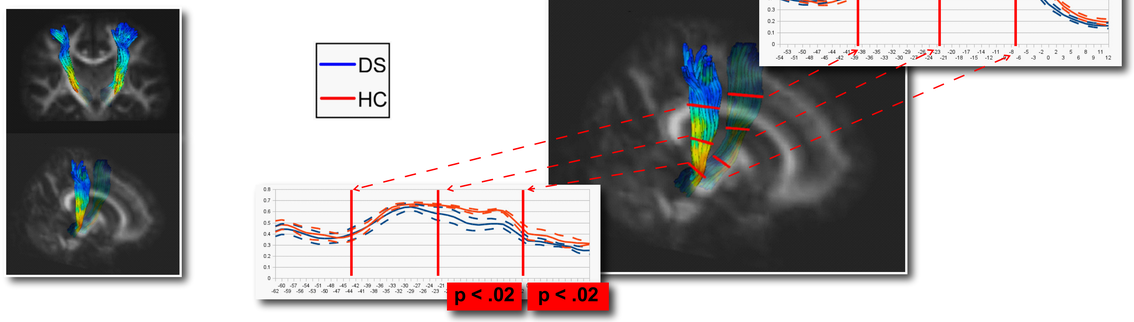
Average volumes of brain tissues, fluid and intra-cranial space for the 2 populations (8 DS and 9 HC).

White matter integrity analysis

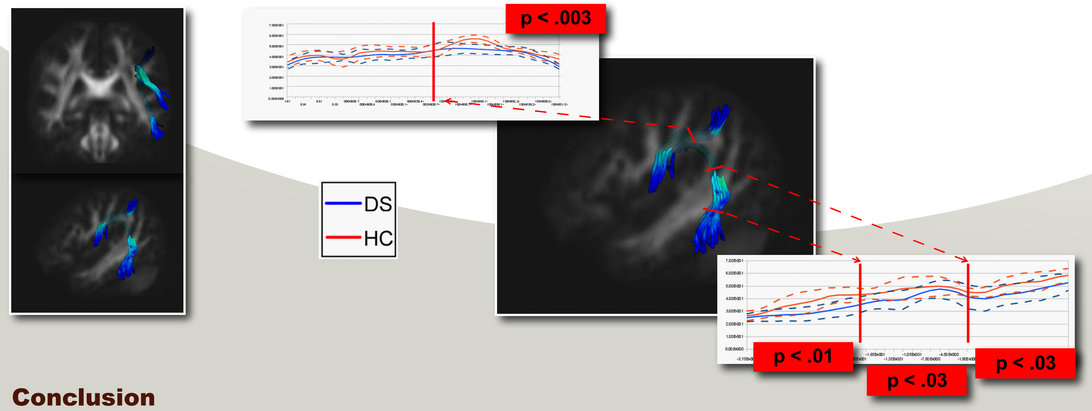
Down syndrome subjects show **cognitive differences** that can be characterized in the white matter integrity of specific brain connectivity pathways. Brain white matter analysis can highlight relationship of genotype and brain connectivity analysis.

► Most portions analyzed tracts show highly significant differences between groups.

Motor tract: associated with motor functions



Arcuate tract: associated with language



Conclusion

Volumetric analysis demonstrates **significant differences of the trajectories** of brain anatomy development in DS subjects → Refined exploration of localized volume and shape differences with help to define new biomarkers of disease.

White matter integrity analysis **reflects the observed cognitive differences** between Down syndrome and control groups → White matter tract analysis might serve as a biomarker of specific aspects of cognitive development.

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