Down syndrome: Bridging Genes, Brain and Cognition

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**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.

**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.