

Erasmus MC



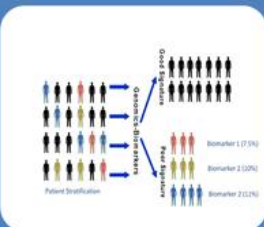
Mission Omics into the Clinic

- Support molecular diagnostic devices
- Enable clinicians to deliver innovative therapies
- Optimize care processes to improve a patients health



Vision Personalized Health Care

- Educate, Develop and Support solutions
- Support collaborative multidisciplinary decisionmaking
- Introduce patient- centric value-based health care



Strategy Patient Stratification

- Molecular Diagnostics
- Targeted Treatment

Data Analysis of Co-morbidities



Health = Genetics + Lifestyle + Environment



Health = Risk + Behaviour + Exposure

Knowledge Mining using AI



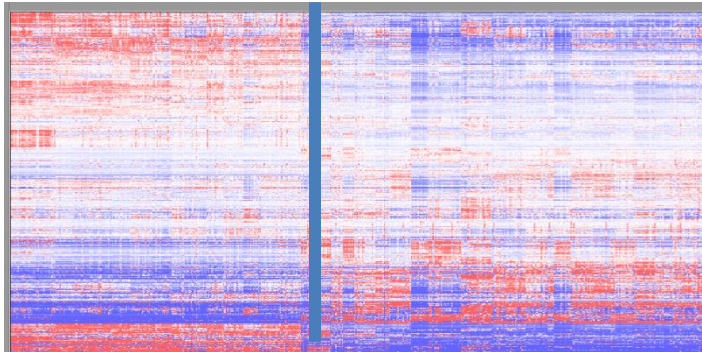
YK YK

Textmining
Literature



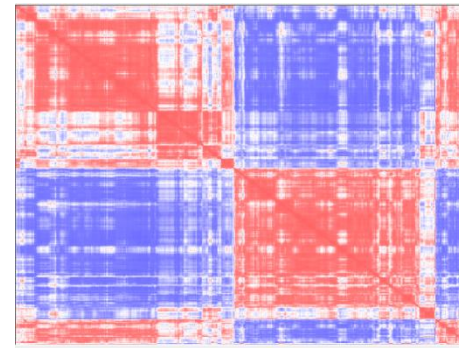
YK DK

Lab Experiment
Diagnostic Test



DK YK

Data Visualization
Population Data

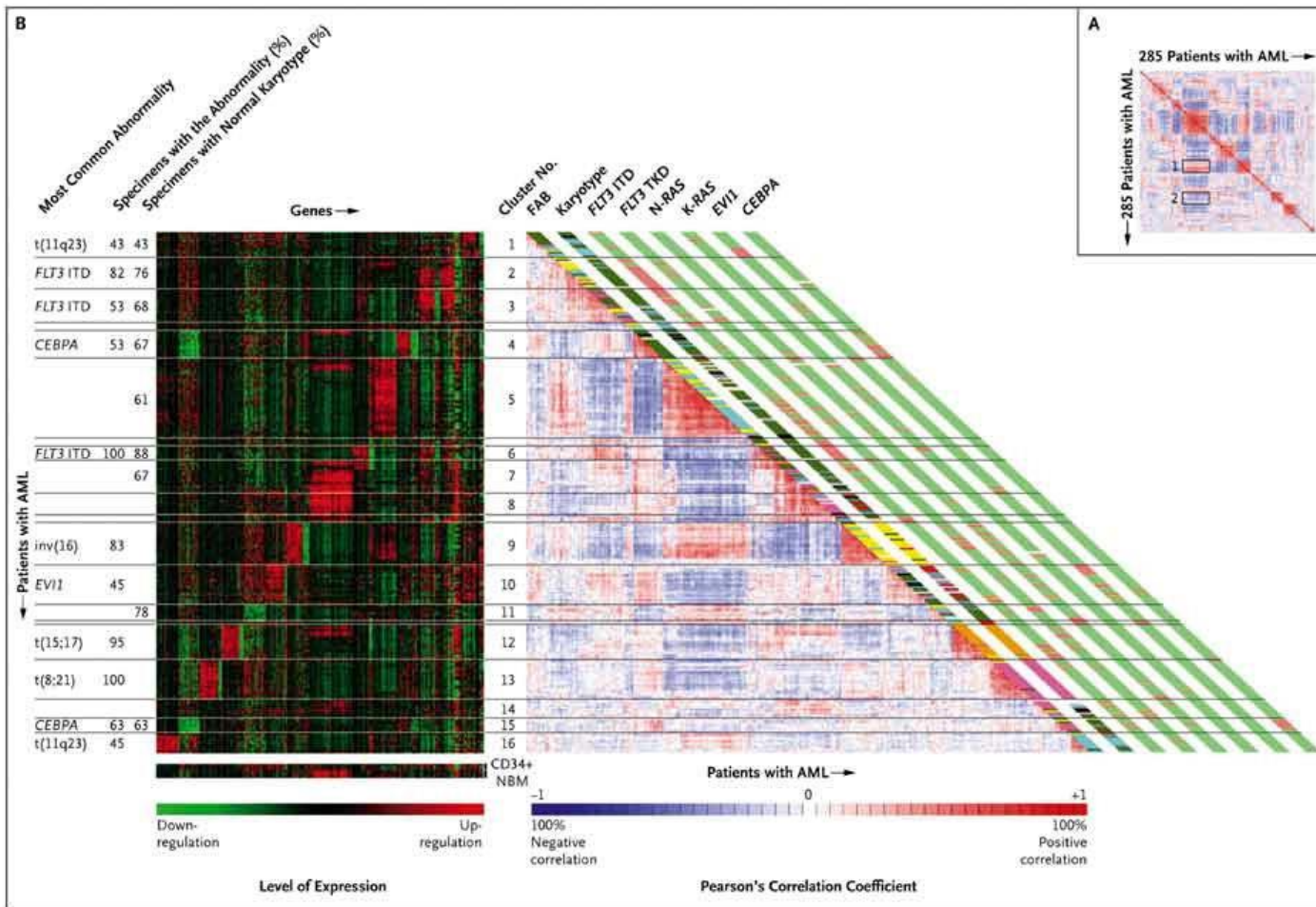


DK DK

Network Analysis
Molecular Pathway

Best Practices in Big Data

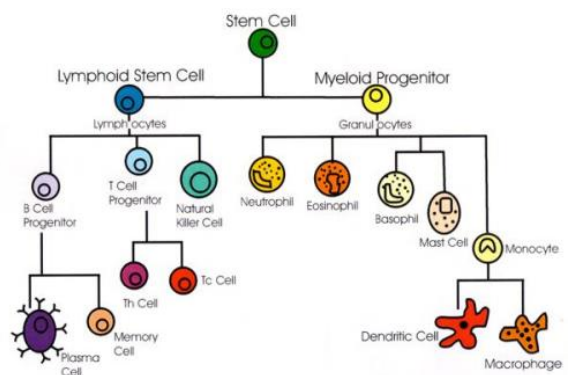
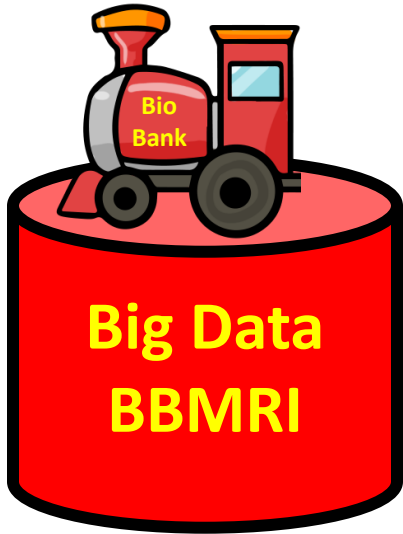
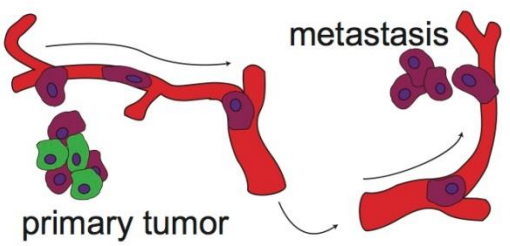
- RNA Expression based AML Patient Stratification -



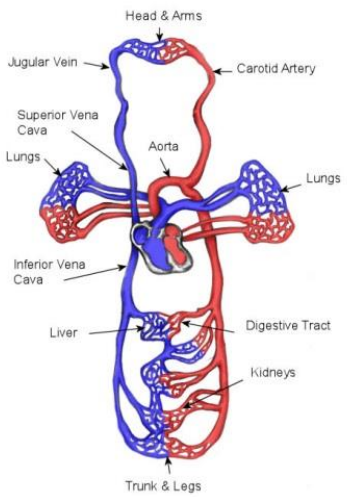
Source: NEJM 2004 Valk et al.

Data Re-use: Need for Shared Storage & AI

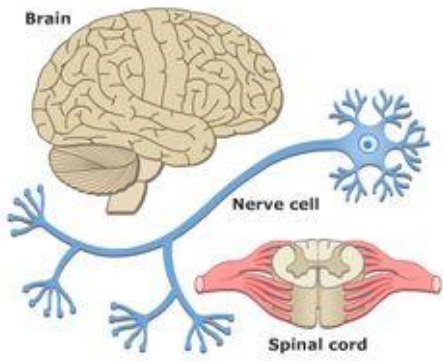
Cancer



Immune & Transplantation



Cardio

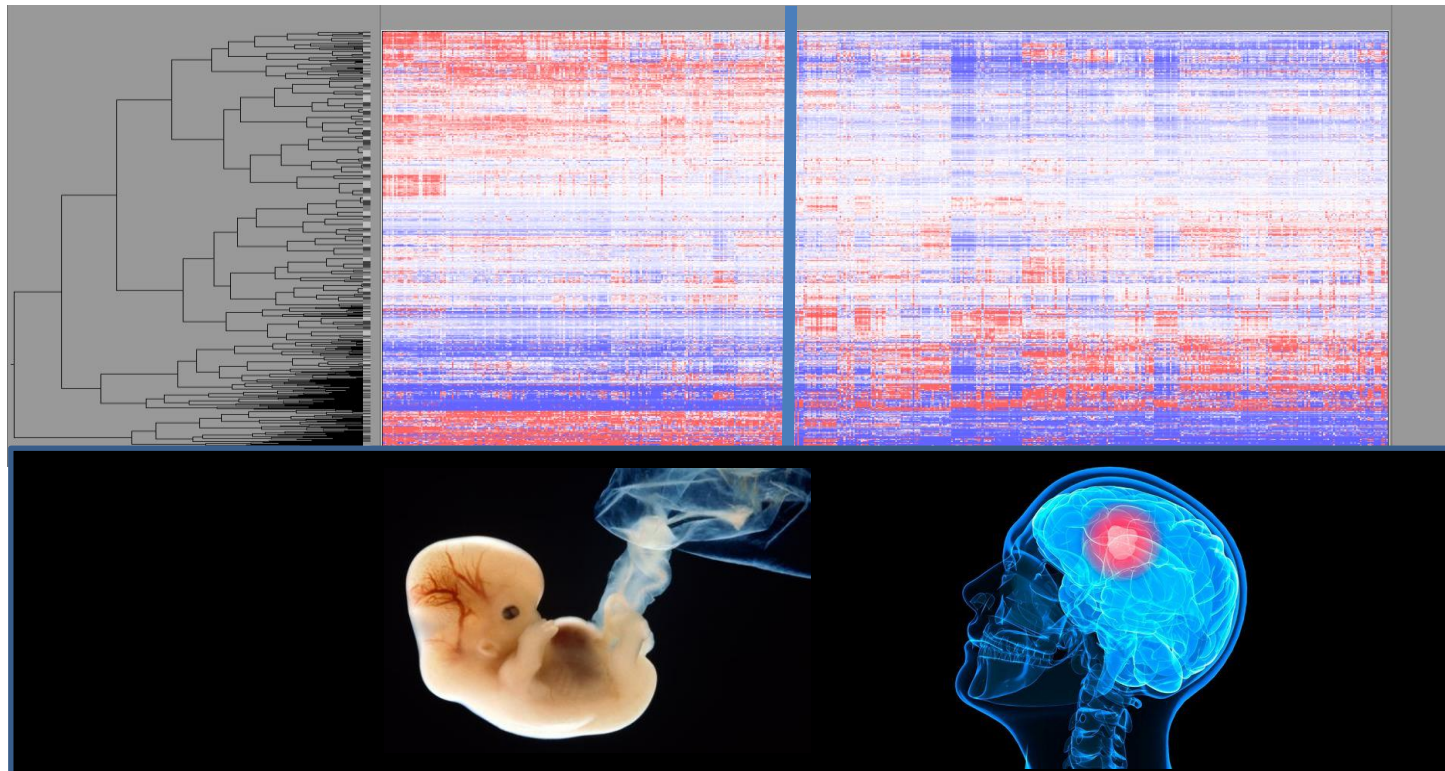
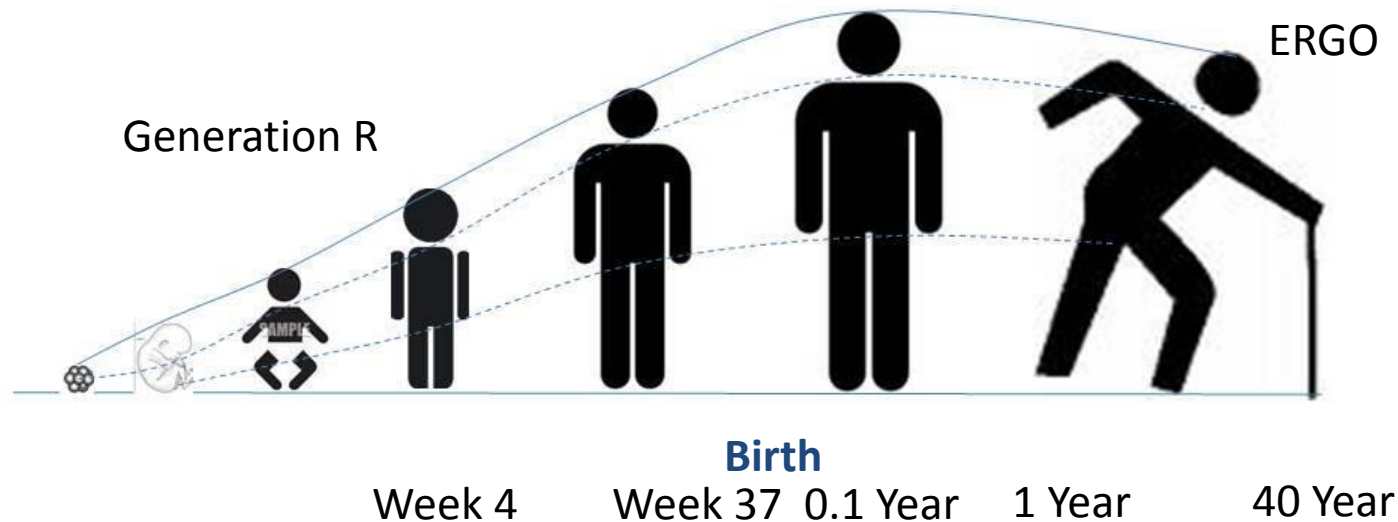


Neuro



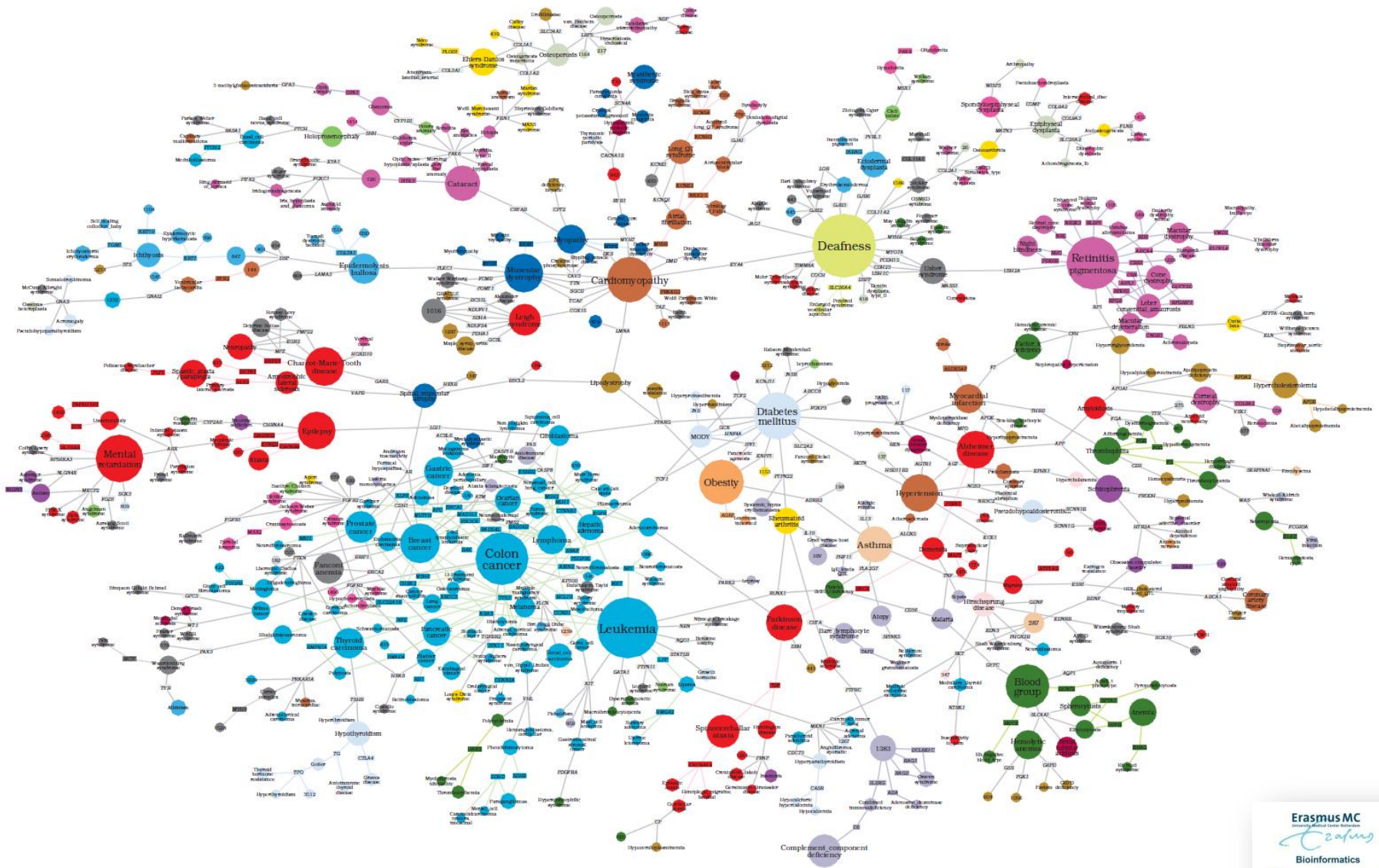
Child

CNS Longitudinal Data



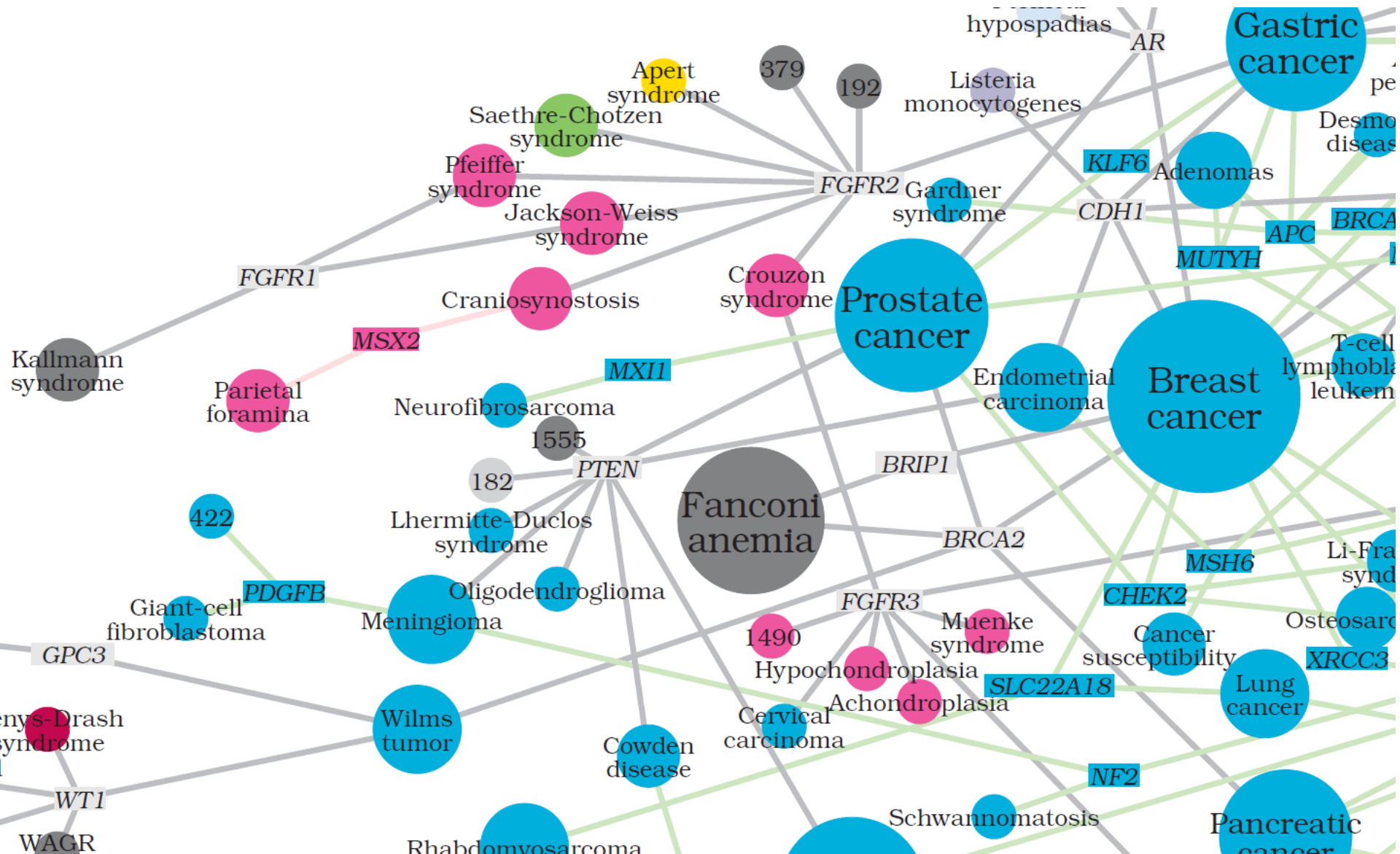
Diseaseome: Connectivity Genes & Disease

- Network of Known Mutations -

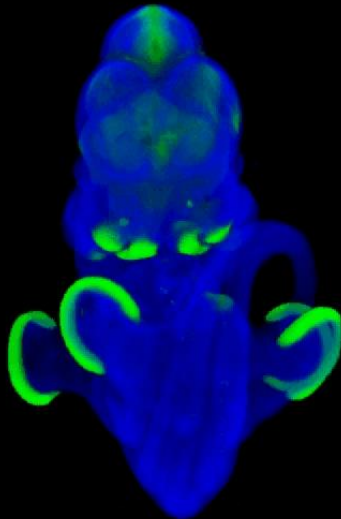
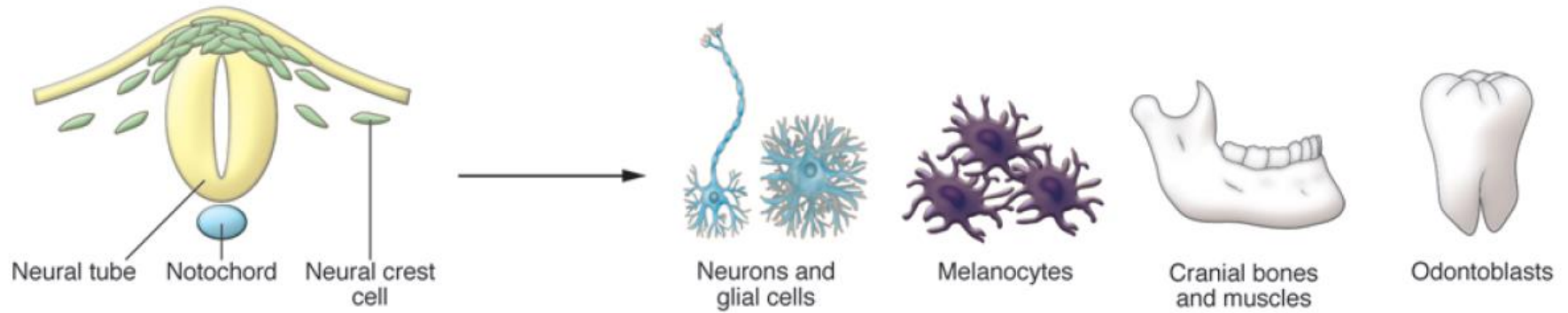


Ref: doi: 10.1073/pnas.0701361104

Mesenchymal Tissue related Disease looks like Cancer Biology



Mutations in Cancer & Development of Craniofacial & Limb structures

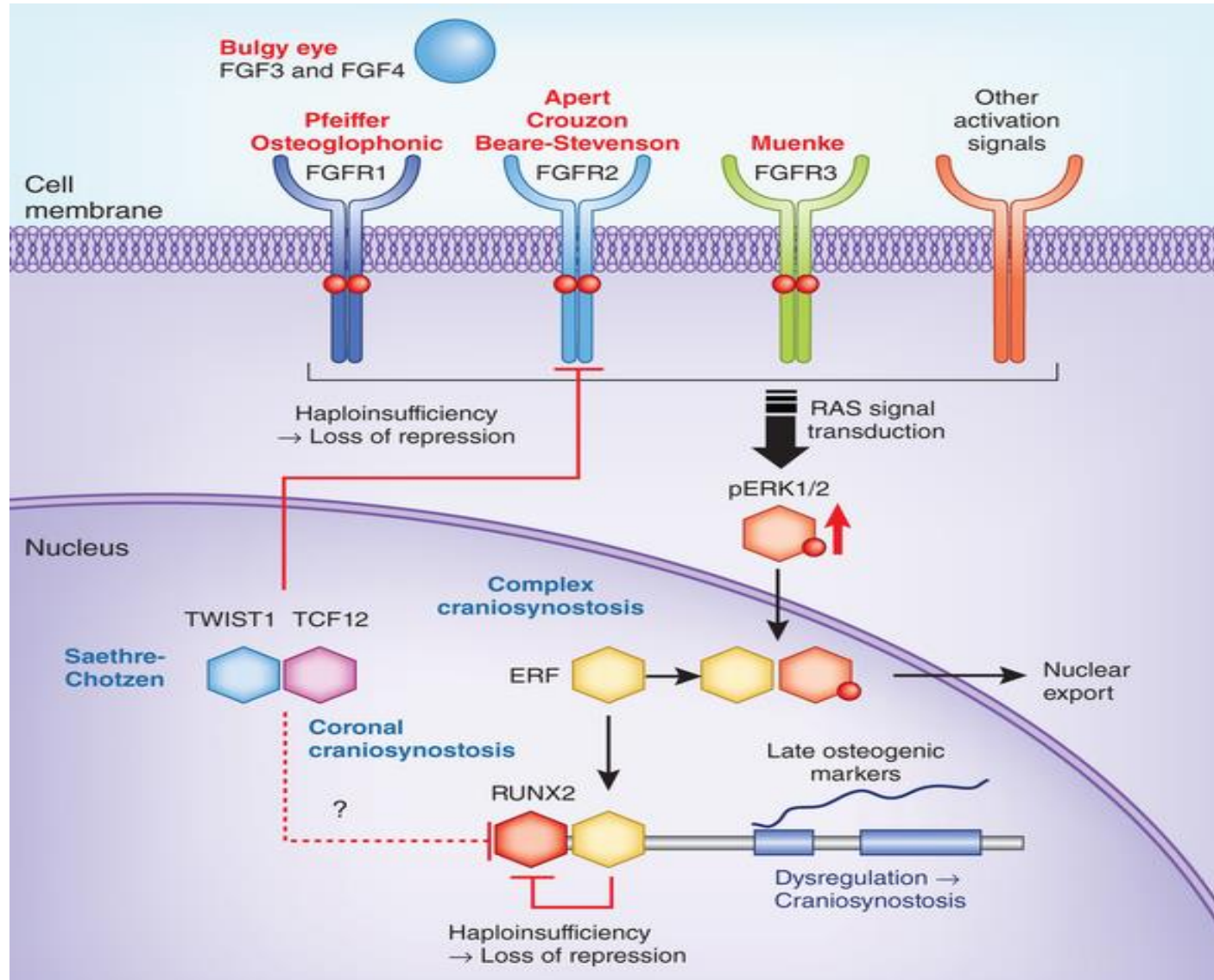


Mouse MSX2 Expression



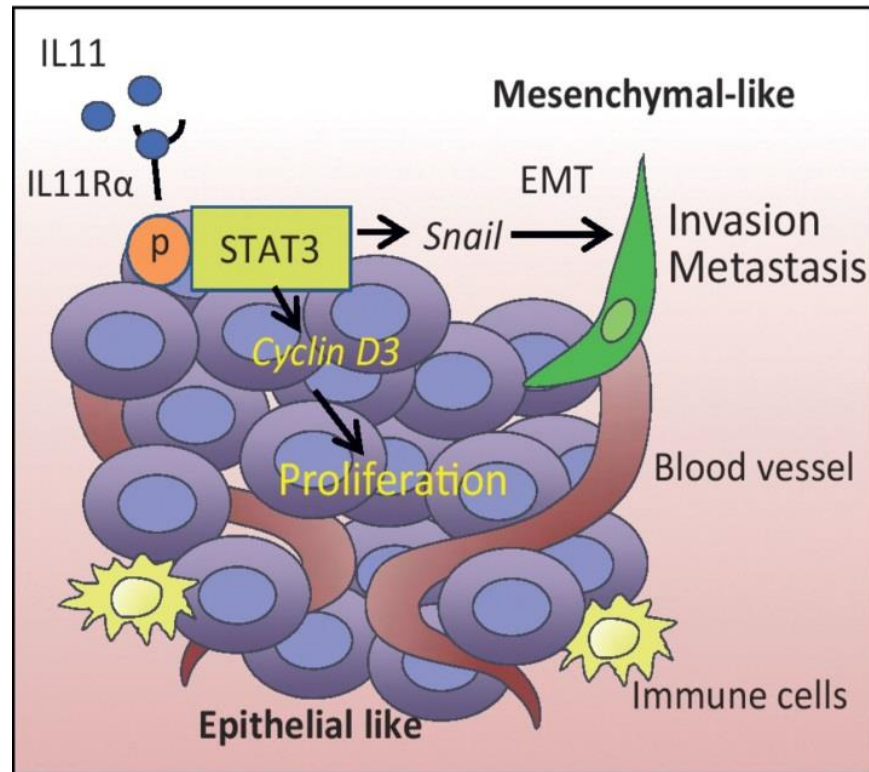
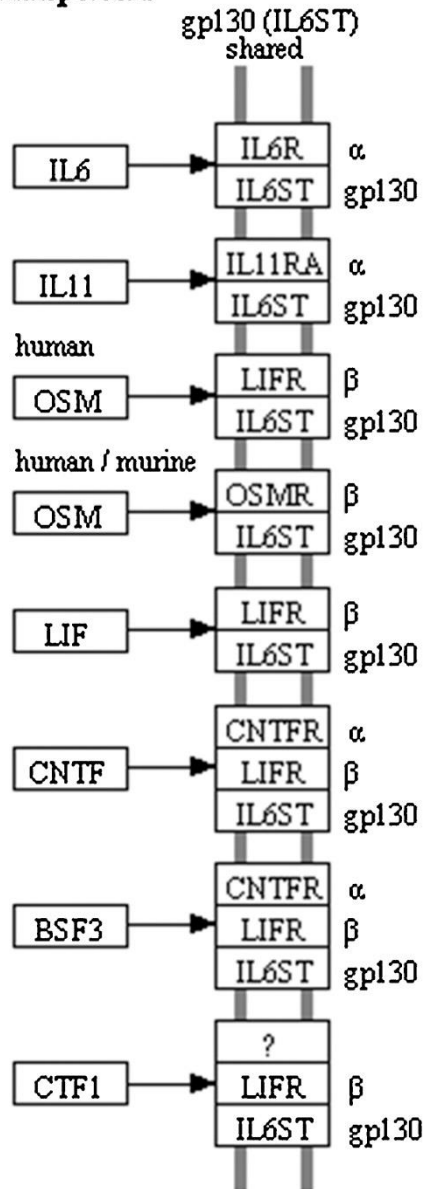
Patient with MSX2 mutation

Syndromes linked to Growth & Apoptosis

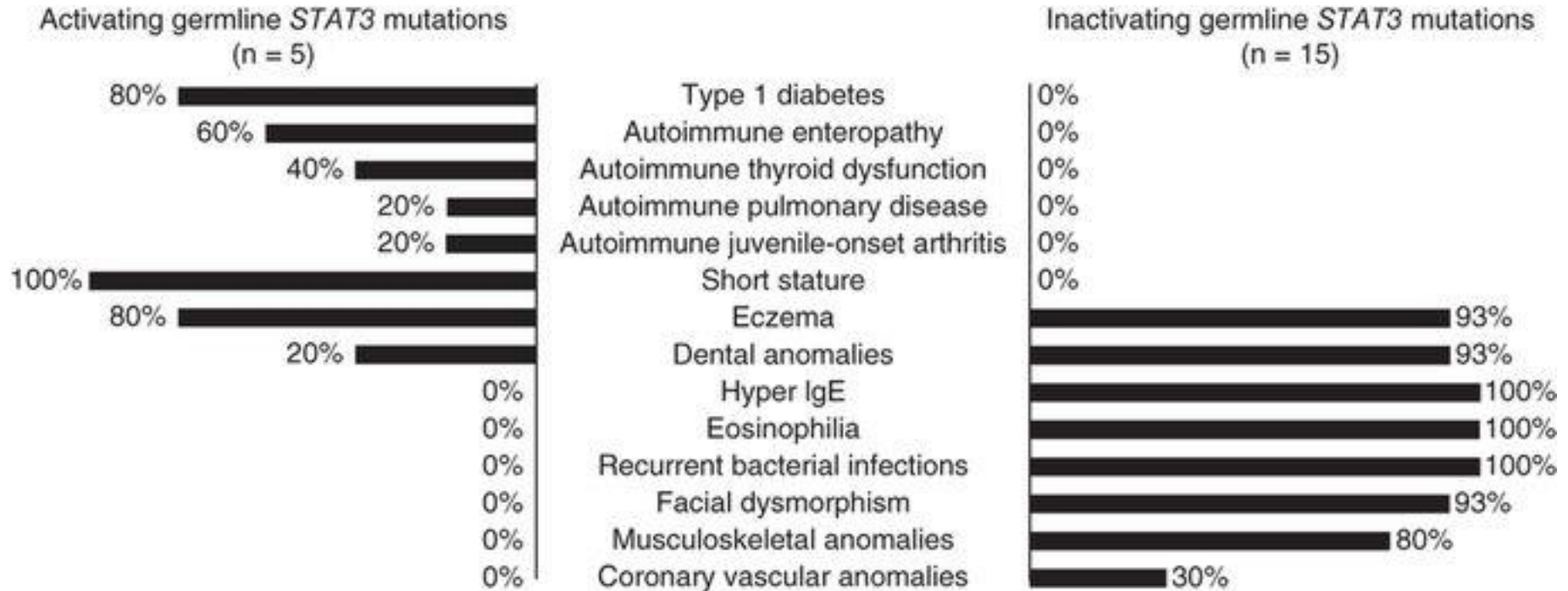


IL-6 is the closest cytokine to IL-11

Hematopoietins



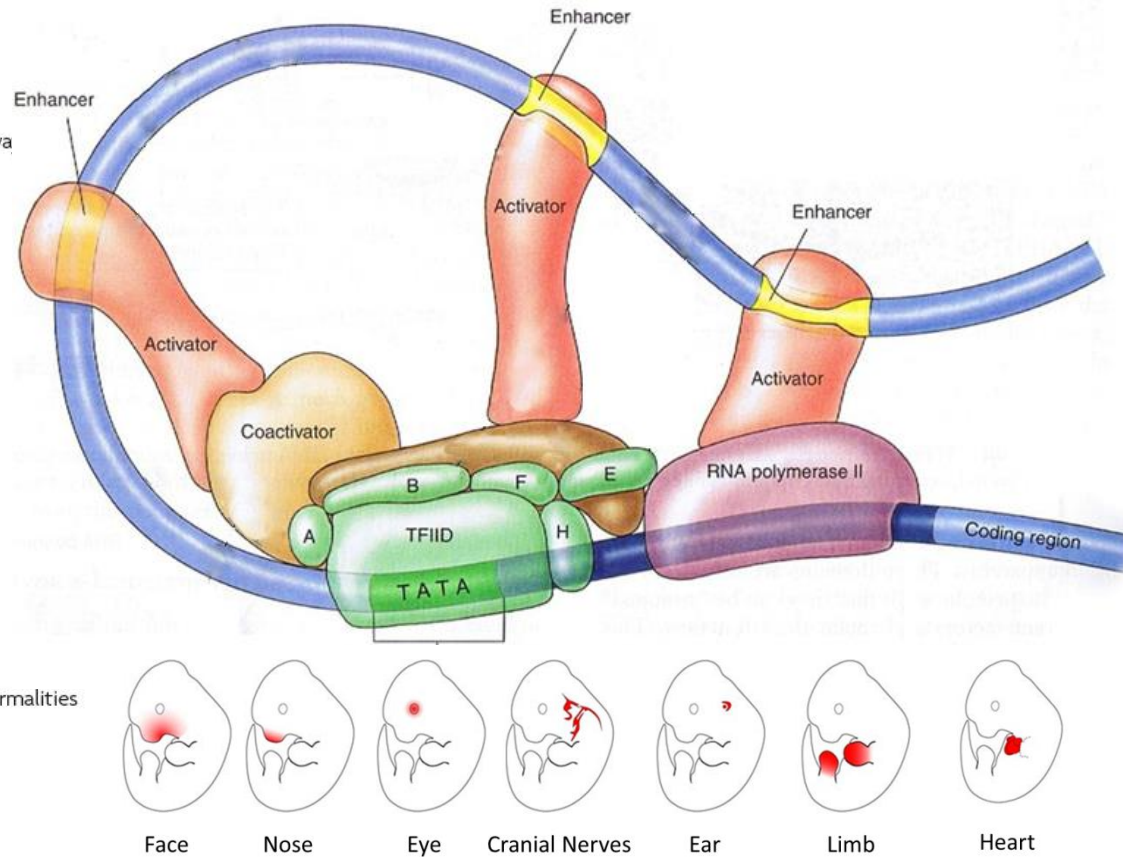
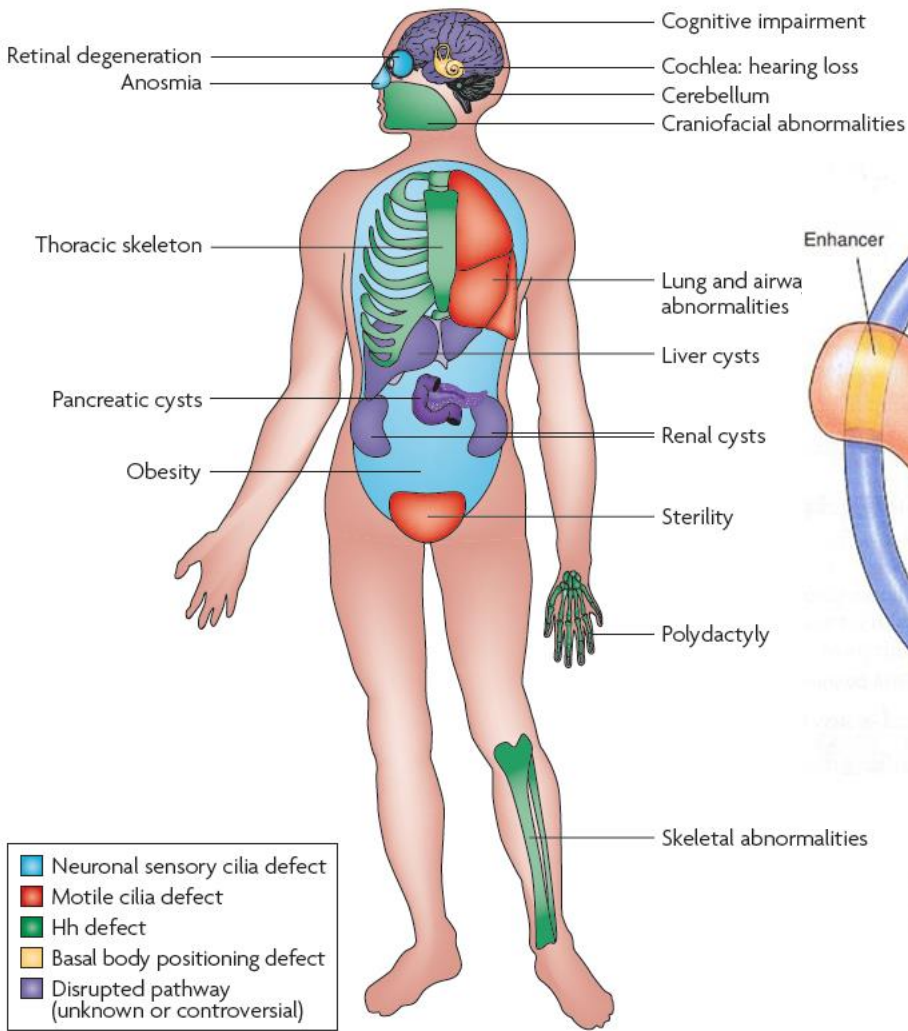
Activating and De-activating STAT3 Mutations



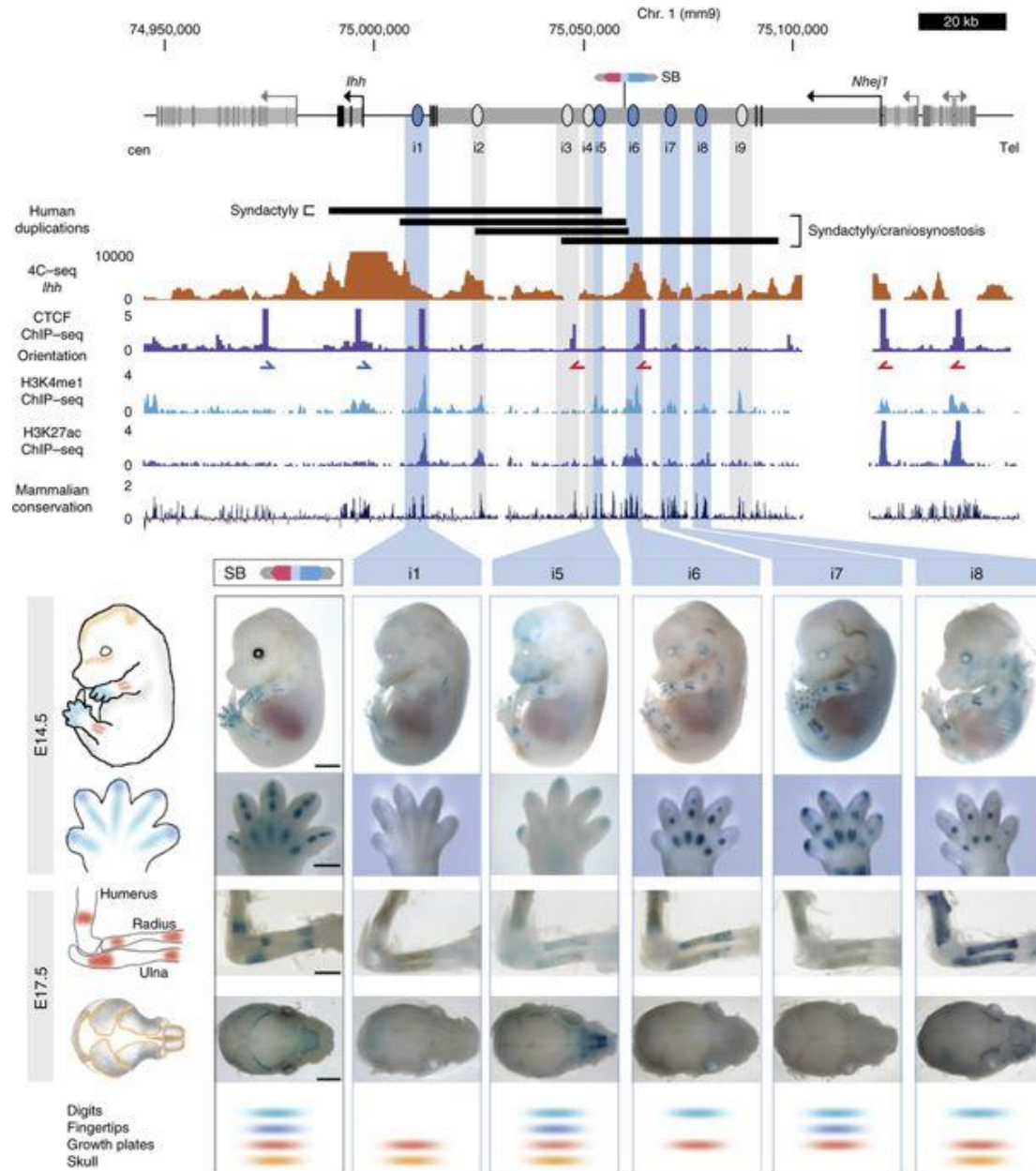
Gain of Function

Loss of Function

Why Whole Genome Sequencing.....?

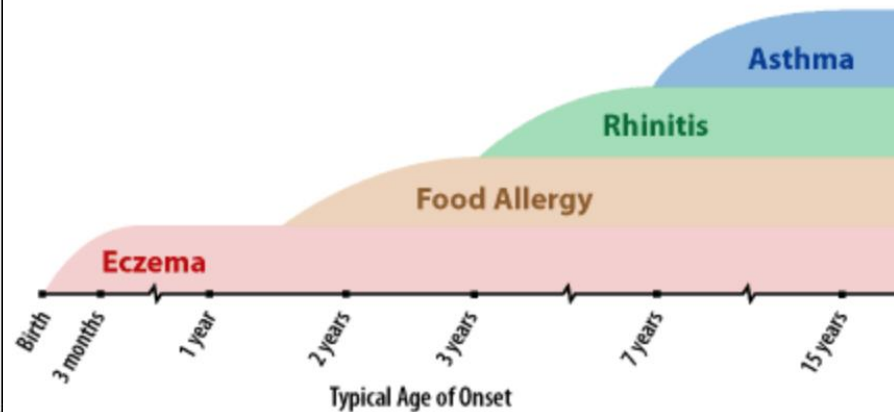
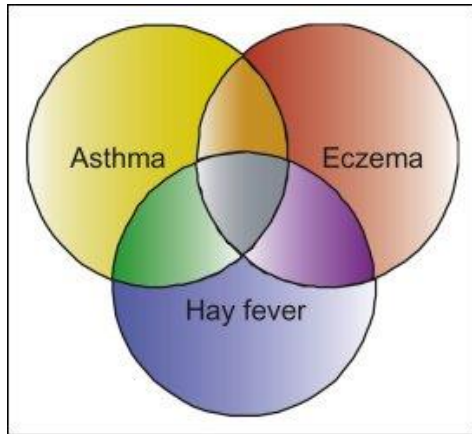


Need for WGS re-use of Genetic Data



Dermatology, Immunology Internal Medicine & Pulmonology

- Longitudinal Data -



The physician identifies a patient who may be appropriate for genetic testing.



The physician requisitions a test and refers the patient to **Consent**



The genetic counselor provides pre-test genetic counseling.



The genetic counselor facilitates genetic testing.



The genetic test results become available.



The genetic counselor provides post-test genetic counseling.



Test



Classify



Interpret



Publish

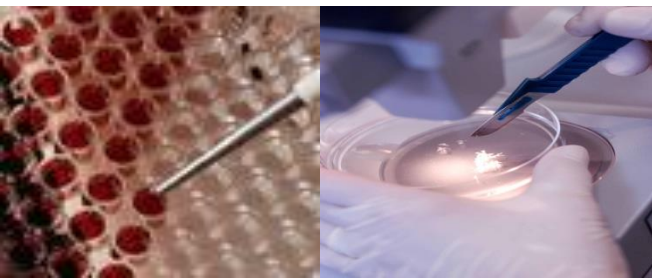


Clinical
Decisions



Take Action!

Precision Medicine / Healthcare Recommendations:



Education

Innovation in teaching program



Reimbursement

Genome First approach (WGS)



Legislation

Open data, Re-use



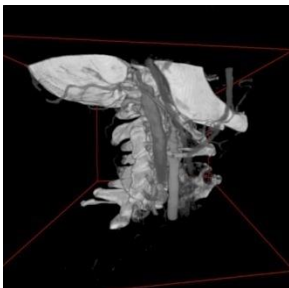
Regulation

GDPR reality check

Conclusion



New opportunities for
disease management



- Expression based classification of biological subgroups
- Identifying DNA variants relevant for Dx & therapy
- Companion diagnostics for directed therapy
- Assessing and managing risk

END