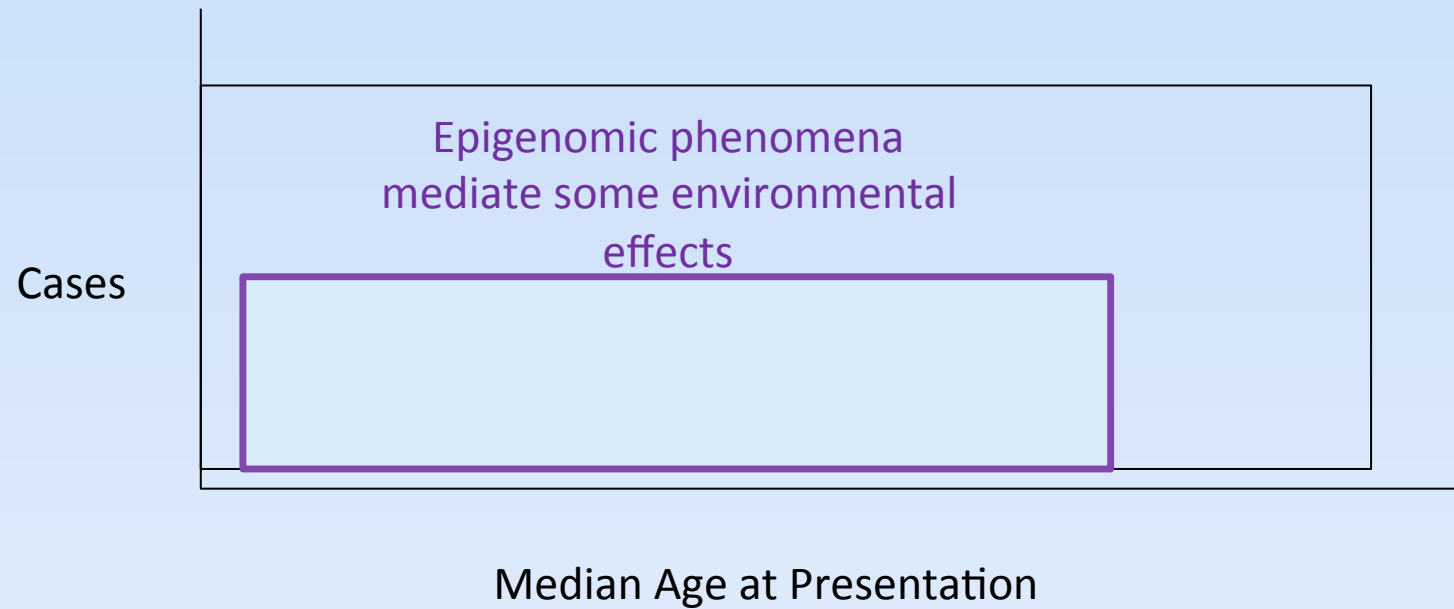
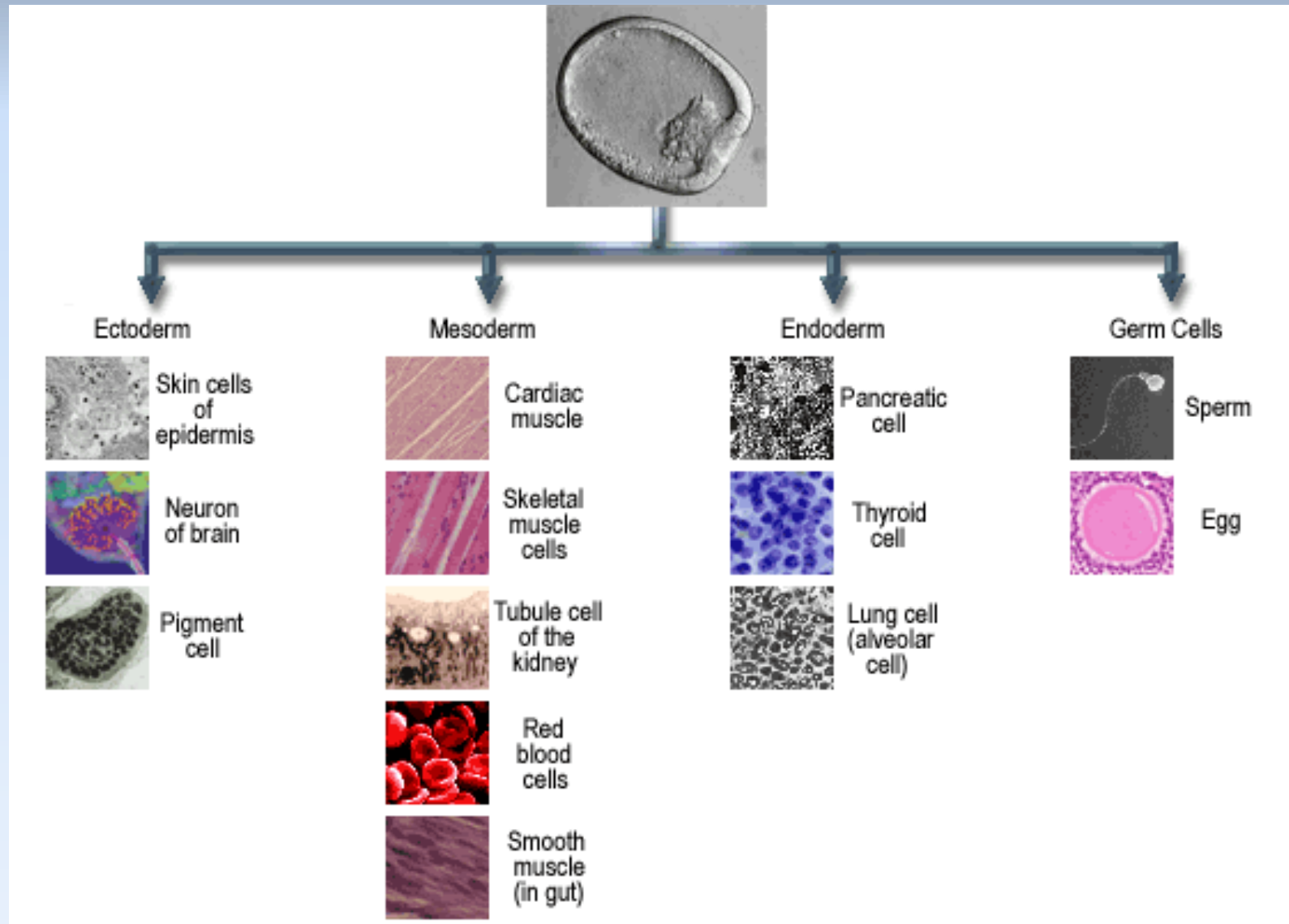


# Rethinking Disease



*A completely hypothetical example to  
illustrate a point*

# How can one cell become so many different things?



## A Few Key Points to Remember

*All the cells in a multicellular organism contain essentially the same DNA*

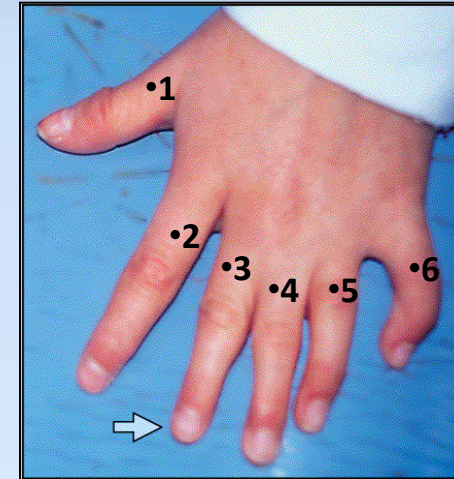
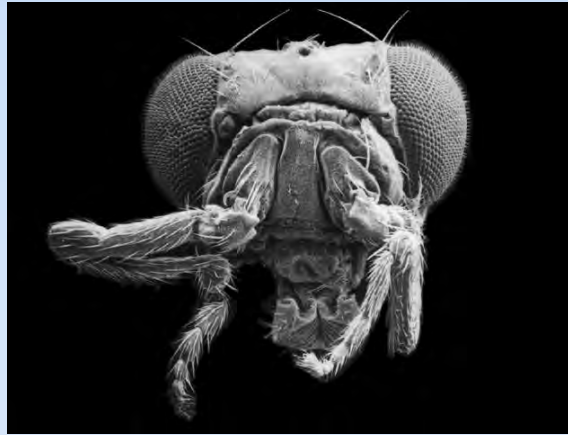
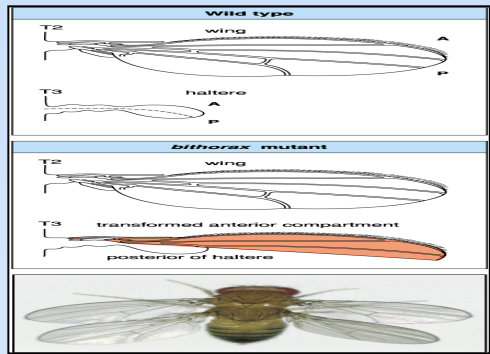
A cell typically expresses only a fraction of its  
Genes

Different types of cells arise because different  
sets of genes are being expressed

Cells can change the pattern of genes they  
express in response to changes in their  
environment

# What happens when gene regulation goes awry?

## Developmental abnormalities (birth defects)



### Disease examples

- Chronic myeloid leukemia
- Rheumatoid arthritis

# NCBI BioProject

The image shows a screenshot of the NCBI BioProject search results page for the query 'tuberculosis'. The page is divided into several sections, with a red box highlighting the left sidebar and the main results area. The sidebar contains a list of filters, including Project Types, Data Types, Project Data, and Organism Groups. The main results area displays a list of search results, with the first result being 'Full genome sequence of Mycobacterium tuberculosis'.

**NCBI BioProject** Search Results for **tuberculosis**

**Project Types**

- Umbrella (29)
- Primary submission (2792)
- RefSeq (153)

**Data Types**

- Clone ends (1)
- Epigenomics (9)
- Genome sequencing (2298)
- Metagenome (1)
- Other (42)
- Proteome (3)
- Targeted locus (3)
- Transcriptome (275)
- Variation (7)

**Project Data**

- Nucleotide (1930)
- Protein (1808)
- Assembly (1927)
- SRA (2045)
- GEO DataSets (280)

**Organism Groups**

- Human (74)
- Bacteria (2627)
- Fungi (1)
- Mammals (124)
- Other vertebrates (5)

**Results: 1 to 20 of 2821**

- Full genome sequence of Mycobacterium tuberculosis**  
Taxonomy: *Mycobacterium tuberculosis*  
Project data type: Other  
Attributes: Scope: Monoisolate; JLU  
Accession: PRJEB4918 ID: 2821
- Mycobacterium tuberculosis**  
Whole genome sequencing strains  
Taxonomy: *Mycobacterium tuberculosis*  
Project data type: Genome sequencing  
Attributes: Scope: Multisolate; M  
Royal Tropical Institute  
Accession: PRJNA285080 ID: 2821
- Gene expression profiling**  
Organism: Homo sapiens  
Taxonomy: *Homo sapiens* (human)  
Project data type: Transcriptome  
Attributes: Scope: Multisolate; M  
Department of Computer Science  
Accession: PRJNA284331 ID: 2821
- Mycobacterium tuberculosis**  
Mycobacterium tuberculosis  
Taxonomy: *Mycobacterium tuberculosis*  
Project data type: Genome sequencing  
Attributes: Scope: Monoisolate; R  
Robert Koch Institute  
Accession: PRJNA284305 ID: 2821

**Project Types**

- Umbrella (29)
- Primary submission (2792)
- RefSeq (153)

**Data Types**

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# NCBI GEO




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GEO help: Mouse over screen elements for information.

Scope:  Format:  Amount:  GEO accession:

**Series GSE48059** [Query DataSets for GSE48059](#)

Status Public on Jul 03, 2014  
 Title Characterization of miRNomes in acute and chronic myeloid leukemia cells  
 Organism [Homo sapiens](#)  
 Experiment type Non-coding RNA profiling by high throughput sequencing  
 Summary An in-depth analysis of miRNomes in 3 human myeloid leukemia cell lines was carried out to comprehensively identify miRNAs that distinguish acute and chronic myeloid leukemias and relate to myeloid cell differentiation.

Overall design Characterization the miRNomes in 3 myeloid leukemia cell lines.

Contributor(s) [Hu S, Fang X](#)  
 Citation(s) Xiong Q, Yang Y, Wang H, Li J et al. Characterization of miRNomes in acute and chronic myeloid leukemia cell lines. *Genomics Proteomics Bioinformatics* 2014 Apr;12(2):79-91. PMID: [24755403](#)

Submission date Jun 18, 2013  
 Last update date Jul 22, 2014  
 Contact name yadong yang  
 E-mail [yangyd@big.ac.cn](mailto:yangyd@big.ac.cn)  
 Organization name Beijing Institute of Genomics  
 Street address No.1-104 Beichen West Road, Chaoyang  
 City Beijing  
 ZIP/Postal code 100101  
 Country China

Platforms (1) [GPL9115](#) Illumina Genome Analyzer II (Homo sapiens)  
 Samples (3) [GSM1167069](#) HL60  
[GSM1167070](#) K562  
[GSM1167071](#) THP1

**Relations**  
 BioProject [PRJNA208839](#)  
 SRA [SRP026144](#)

Download family	Format
<a href="#">SOFT formatted family file(s)</a>	<a href="#">SOFT</a> <a href="#">?</a>
<a href="#">MINIML formatted family file(s)</a>	<a href="#">MINIML</a> <a href="#">?</a>
<a href="#">Series Matrix File(s)</a>	<a href="#">TXT</a> <a href="#">?</a>

Supplementary file	Size	Download	File type/resource
<a href="#">GSE48059_3cell_cf_10e5.txt.gz</a>	10.0 Kb	<a href="#">(ftp)</a> <a href="#">(http)</a>	TXT
<a href="#">SRP/SRP026/SRP026144</a>		<a href="#">(ftp)</a>	SRA Study

Processed data is available on Series record  
 Raw data provided as supplementary file



*Gene-Environment Interactions are no Longer a Black Box,  
but we are Going to Need Lots of Data to  
Translate this into Medicine!*

Approximate samples needed:

- Rare genetic variants ~ Hundreds
- More common genetic variants ~ Thousands
- Genetic variation-environment interplay ~ Millions

**The most efficient way to do this is to share  
and reuse data in a public forum.**

# For more information go to:

[ncbi.nlm.nih.gov/learn](http://ncbi.nlm.nih.gov/learn)


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### UPCOMING EVENTS

**How to upload and analyze dbGaP data in the Cloud**  
FEBRUARY 3, 2016  
Online Webinar: 1:00-2:00pm





**Five ways to submit next-gen sequence data to NCBI's Sequence Read Archive**  
FEBRUARY 17, 2016  
Online Webinar: 1:00-2:00pm

**"NCBI Resources for Patent Searchers" at the PIUG Biotechnology 2016 Conference**  
FEBRUARY 24, 2016  
Workshop

**A Librarian's Guide to NCBI**  
MARCH 7-11, 2016  
Workshop

**Experimental Biology 2016 Annual Meeting**  
APRIL 2-6, 2016  
Conference

**"Practical Bioinformatics for the Clinic" at the NLM Biomedical Informatics Course**  
APRIL 8, 2016  
Presentation

Webinars & Courses	Conferences & Presentations	Tutorials	Documentation
In-person courses, live webinars and webinar recordings	Booth exhibits and workshops at scientific conferences	Tutorials: Training materials in HTML, PDF and video formats	Online manuals, handbooks, fact sheets and FAQs
			

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