

# Ohno's Evolution by Duplication and DNA Correlations

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# Outline of the talk

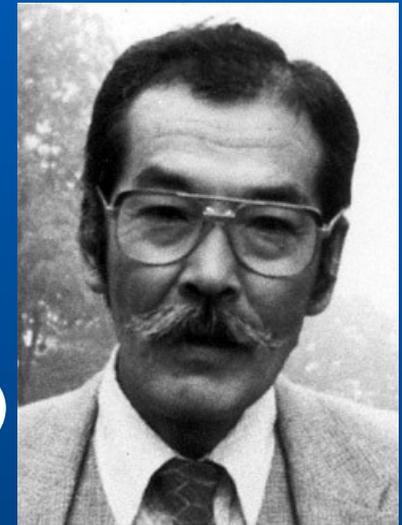
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- Ohno's evolution by gene duplication
- Duplication-mutation model (expansion-modification model)
- Confirmation of  $1/f$  noise in DNA sequences as predicted by the duplication-mutation model
- Duplication in bacteria genomes?
- Music as the second example where the three themes converge ( $1/f$  noise - redundancy in musical composition - duplication-mutation model)

# Susumu Ohno (1928-2000)

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- Undergraduate degree in veterinary (1949)
- Graduate degree in immunology (1953)
- UCLA, then City of Hope National Medical Center (50s-retired)
- One X-chromosome being heterochromatic(1959)
- Evolution by gene duplication (1970)
- Term “junk DNA” (1972)
- Relating DNA with music (1986)



## From *Evolution by Gene Duplication* (1970)

*“Had evolution been entirely dependent upon natural selection, from a bacterium only numerous forms of bacteria would have emerged. The creation of metazoans, vertebrates, and finally mammals from unicellular organisms would have been quite impossible, for such big leaps in evolution required the creation of new gene loci with previously nonexistent function. Only the cistron [gene] that became redundant was able to escape from the relentless pressure of natural selection. By escaping, it accumulated formerly forbidden mutations to emerge as a new gene locus.”*

“Natural selection merely  
modified while  
redundancy created”

-S. Ohno

## Pre-1970 works on gene duplication

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- Kuwada (1911), Tischler (1915)
- **JBS Haldane (1932)**, *The Causes of Evolution* (Harper and Bros)
- Bridges's observation of gene duplication in *Drosophila* (1935-36)
- **Serebrovsky (1938)**: selection is relaxed in genes that duplicate
- Muntzing (1936), Tischler (1935), Nishiyama (1934)
- **Gulick (1944)** "increases in gene count.. (to) great complexity"
- Goldschmit (1940), Metz(1947), Huxley (1942): **importance of..**
- **SG Stephens (1951)**, "Possible significance of duplication in evolution", *Adv. Genetics*, 4:247-265.
- **Lewis (1951)** "pseudoallelism & gene evolution" **linked duplicates**
- S Ohno, U Wolf, NB Atkin (1968), "Evolution from fish to mammals by gene duplication", *Hereditas*, 59:169-187.

See **Taylor & Raes (2004)**

## Some examples of disease genes

BRCA1	Breast cancer	1
BRCA2	Breast cancer	1
CFTR	Cystic fibrosis	15
RPGR	Retinitis pigmentosa	1
PTPN22	Many autoimmune diseases	3
PSEN	Alzheimer's disease	2
WRN	Werner syndrome	4
HD	Huntington's disease	1
PRKN	Juvenile Parkinson	1
CSTB	Progressive myoclonus epilepsy	2

the general principle of Ohno's duplication is true, but...

- **Whole genome duplication (polyploidization) or regional/local/segmental duplication?**  
[polyploids tend to be unstable]
- **Duplication of genes or duplication of a piece of DNA with possibly no functions?**  
[then “gene” duplication is a by-product instead of a design]
- **Is redundancy caused by duplication the only source of robustness against mutations?**  
[system robustness is another source]

## DNA-Music connection: I

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- **DNA sequences all exhibit  $1/f$  spectra (no exception yet)**
- **Musical time series all exhibit  $1/f$  noise (both loudness and pitch, both musical signal and speech)**

Voss & Clarke (1975) “ $1/f$  noise in music and speech”, *Nature*, 258:317-318.

## **DNA-Music connection: II**

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- **DNA sequence is redundant (full of repeats)**
- **Musical series is also redundant (repetitive)**

## DNA-Music connection: III

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- **The generation/elongation of DNA sequences are driven mainly by duplication in various form (genomewide, segmental,...)**
- **The musical composition process consists of re-usage of the main/minor themes**
- **Can both be modeled by some form of duplication-mutation models?**

“We have formerly seen that parts many times repeated are eminently liable to vary in number and structure; consequently it is quite probable that natural selection, during the long-continued course of modification, should have seized on a certain number of the primordially similar elements, many times repeated, and have adapted them to the most diverse purposes.

Charles Darwin, 1859 (*The Origin of Species*, page 477)