Genetic stories behind village sign languages
– the co-evolution of deafness with sign language –

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Overview

- Genetic mechanisms in hearing loss
- *DFNB1* (ABSL) and *DFNB3* (Kata Kolok)
- Models of genetic hearing loss and sign language
- The origins, evolution and stability of village sign languages
Genetic mechanisms in hearing loss

- Many types of hearing loss

- Acquired
  - Drugs
  - Infections
  - Head injury
  - Noise
  - Aging

- Congenital
  - Prenatal infections
  - Genetics
• Many types of hearing loss → **genetic role**
Some examples: TECTA

- Ear Canal
- Inner Ear
- Eustachian Tube
- Pinna
- Middle Ear
- Eardrum

The Organ of Corti

TECTA
Some examples: TECTA

- Tectorial membrane:
  → collagen
Some examples: \textit{TECTA}

- Tectorial membrane:
  - \rightarrow collagen
  - \rightarrow non-collagenous proteins
    - \rightarrow \textit{\textalpha-tectorin}

\textit{TECTA} gene (11q23.3)
Some examples: **TECTA**

- **Tectorial membrane:**
  - → collagen
  - → non-collagenous proteins
    - → *α-tectorin*

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TECTA gene

$DFNA12$ - dominant
Some examples: TECTA

- Tectorial membrane:
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TECTA gene

→ DFNA12 - dominant
→ DFNB21 - recessive
Some examples: mitochondrial 12S rRNA
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Own genetic code
→ own translation machinery
→ own ribosomes

Figure 1

Small subunit ribosomal RNA

MTRNR1
Some examples: mitochondrial 12S rRNA

Mitochondria → ancient bacteria (endosymbiotic theory)
Some examples: mitochondrial 12S rRNA

Mitochondria → ancient bacteria (endosymbiotic theory)
→ mitochondrial ribosomes are very similar to bacterial ribosomes

Mutations (e.g. A1555G)

even more similar

modifiers
Some examples: mitochondrial 12S rRNA

Hair cells are more affected by mitochondrial ribosomal impaired function more than other cells? → specific phenotype

Mutations (e.g. A1555G)

even more similar

modifiers
Congenital non-syndromic deafness

- Many loci (e.g., http://hereditaryhearingloss.org/main.aspx?c=.HHH&n=86163)
- Autosomal dominant: $DFNAnn$ (~25)
Congenital non-syndromic deafness

- Many loci (e.g., http://hereditaryhearingloss.org/main.aspx?c=.HHH&n=86163)

- Autosomal dominant: $DFNAnn$, recessive: $DFNBnn$ (~40)
DFNB1A/B (GJB2 and GJB6)

- Autosomal recessive
- 13q12.11
- **GJB2** (Gap junction beta-2; Connexin 26) and **GJB6** (Gap junction beta-6; Connexin 30)
• Autosomal recessive

• 13q12.11

• \textit{GJB2} (Gap junction beta-2; Connexin 26) and \textit{GJB6} (Gap junction beta-6; Connexin 30)

• \textit{GJB2}: \(\sim\)90 mutations \(\rightarrow\) non-syndromic deafness
  – other mutations: syndromes (skin + deafness)

• \textit{GJB6}: some mutations \(\rightarrow\) non-syndromic deafness
  – other mutations: skin disorders

\(\rightarrow\) potassium levels in the inner ear?
DFNB3 (MYO15A)

- Autosomal recessive
- 17p11.2
- MYO15A (unconventional myosin-15; myosin XVa)
  $\rightarrow$ stereocilia

The Organ of Corti
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DFNB3 (MYO15A)

- Autosomal recessive
- 17p11.2
- *MYO15A* (unconventional myosin-15; myosin XVa)
  - → sterocilia
  - → A2674T mutation → non-functional protein → abnormal stereocilia → hearing loss
ABSL: population & evolutionary genetics

- Al-Sayyid Bedouin community
- ~200 years ago
- 3rd generation
- ~3,500 members
- ~3.3% deaf members
- social integration
- ABSL – L2 for hearers
Kata Kolok: population & evolutionary genetics

- Bengkala village, Bali
- ~150-300(?) years ago
- ~10-20(?) generations
- ~2,200 members
- ~2.2% deaf
- social integration
- KK – L2 for hearers
ABSL & Kata Kolok

- **Recessive** → hard to predict
• Recessive → hard to predict
• Non-syndromic → no other ill effects
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• Non-syndromic → no other ill effects
• Inbreeding → increasing frequency
ABSL & Kata Kolok

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- Social integration → increased biological fitness
• Recessive $\rightarrow$ hard to predict
• Non-syndromic $\rightarrow$ no other ill effects
• Inbreeding $\rightarrow$ increasing frequency
• Social integration $\rightarrow$ increased biological fitness

True gene-culture co-evolution
ABSL & Kata Kolok

What can we say about these parameters?

Emergent generalized sign language

Increased biological fitness of deafness mutation

Recessive deafness
long-term
high frequency

Emergent generalized sign language
Models of sign language genetics

- Gene-culture co-evolution tradition
- Genetic epidemiology approach
- Critique and perspective

→ see tomorrow Alessandro Gialluisi's talk
Series of papers:


Gene-culture co-evolution

• Basic ideas:
  – **Sign language**:
    • *monolithic* cultural trait
    • an individual has it or not
    • transmission: *vertical*, oblique, horizontal
    • learning:
      – both parents **sign** → prob. \( b \) and \( c \)
      – single parent signs → reduced by \( l \)
      – oblique: \( f \) and \( g \)
      – horizontal: \( h \) and \( i \)
      – maternal grandparents...
Gene-culture co-evolution

Basic ideas:

- Genetics:
  - diploid biallelic locus
  - genotypes \(AA\), \(Aa\) and \(aa\)
  - \(A\) dominant
  - \(aa\) → deaf from birth
  - allele frequencies: \(p\) (\(A\)) and \(q\) (\(a\))
  - fraction of assortative matings: \(m\)
    - by deafness
    - by sign language use
Gene-culture co-evolution

- Other assumptions:
  - equilibrium
  - $0 < p, q < 1$ (later, $q << 1$)
  - $0 \leq m < 1$
  - $0 \leq b \leq c \leq 1$
  - $0 < l \leq 1$
Gene-culture co-evolution

- **Conclusions:**
  - If sign language “cannot jump a generation”:
    - Assortment by deafness → helps persistence
    - Assortment by signing → hinders persistence
    - Dominant helps more than recessive
    - Horizontal & vertical → don't seem to matter
  - **Grandparental transmission** (language “can jump”):
    - Assortative mating high → grandparental transmission helps
Gene-culture co-evolution

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  → **assortative mating** by deafness helps most!
Genetic epidemiology

• Series of papers:


Genetic epidemiology

- Basic ideas:
  - **Assortative mating** by deafness
  - **Relaxed selection** against deafness
Genetic epidemiology

- Compare E.A. Fay's 19th century data on deafness
- With 21st century data from a comparable sample
Genetic epidemiology

- Compare E.A. Fay's 19th century data on deafness
- With 21st century data from a comparable sample

→ dramatic increase (~5 times) over 100 years
→ linear increase over the last 60 years
→ not assortative mating for deafness per se but linguistic homogamy
Genetic epidemiology

- **Computer simulations:**
  - Agent-based, non-overlapping generations
  - 3 recessive loci
  - Deafness fitness, $f$
  - Assortative mating proportion, $m$
  - Fixed population size (200,000 agents)
  - Fixed sex ratio (1:1)
Genetic epidemiology

\[ f = 0.0 \quad m = 0\% \quad f: 0 \rightarrow 1 \quad M: 0\% \rightarrow 90\% \quad f = 1.0 \quad m = 90\% \]
Genetic epidemiology

Frequent gene

Infrequent genes:
- linked to Cx
- unlinked to Cx

\[ f = 0.0 \]
\[ m = 0\% \]
\[ f: 0 \rightarrow 1 \]
\[ M: 0\% \rightarrow 90\% \]
\[ f = 1.0 \]
\[ m = 90\% \]
Genetic epidemiology

• **Conclusions:**
  - Assortative mating + relaxed selection → increase frequency
  - Linguistic homogamy
  - Initially higher freq. genes → disproportionately amplified
Conclusions:
- Assortative mating + relaxed selection → increase frequency
- Linguistic homogamy
- Initially higher genes disproportionately amplified

→ assortative mating for sign language/linguistic homogamy
The origins, evolution and stability of village sign languages

- Conditions & models → Alessandro

- Observation 1: many potential genes → why do so few get “used” ($DFNB1$, $DFNB3$...)?

- Observation 2: Endogamy/inbreeding is not infrequent → why so few village sign languages?

- Observation 3: Most seem relatively young → where are the old ones?
The origins, evolution and stability of village sign languages

→ probably the village sign languages are **fragile**:  
  - **genetic** conditions (inbreeding, assortment)  
  - **cultural** conditions (social acceptance)  
  - **linguistic** conditions (“critical mass”)

→ most appear, flicker and disappear  
→ **special conditions** for their long-term maintenance  
→ if they are good models for language origins & evolution then maybe language appeared multiple times and died out
Thank you!

Questions?

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