### Asymptotic Behaviour of Homozygosity

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AMS Fall Central Sectional Meeting, Ann Arbor, Michigan

October 20-21, 2018

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## Outline

#### Homozygosity

- Definition
- LLN and Gaussian Limit

#### 2 Large Deviations

- Deviation From Zero
- Deviation From One
- Link Between the Two Rate Functions

#### 3 Main Steps of Proof

## Definition

Let  $\gamma(t)$  denote the gamma subordinator with Lévy measure

$$\Lambda(dx) = x^{-1}e^{-x}dx, x > 0.$$

For any  $\theta > 0$ , let  $J_1(\theta) \ge J_2(\theta) \ge \cdots$  denote the jump sizes of  $\gamma(t)$  over the interval  $[0, \theta]$  in descending order. If we set  $P_i(\theta) = J_i(\theta)/\gamma(\theta), i \ge 1$ , then the law of

$$\mathsf{P}(\theta) = (\mathsf{P}_1(\theta), \mathsf{P}_2(\theta), \ldots)$$

is Kingman's Poisson-Dirichlet distribution  $PD(\theta)$ . It is a probability on the infinite-dimensional simplex

$$abla_{\infty} = \{ \mathbf{p} = (\mathbf{p}_1, \mathbf{p}_2, \ldots) : \mathbf{p}_1 \ge \mathbf{p}_2 \ge \cdots \ge 0, \sum_{i=1}^{\infty} \mathbf{p}_i \le 1 \}.$$

For any integer  $\mathbf{m} \ge 2$ , the function

$$\mathsf{H}(\mathsf{p};\mathsf{m}) = \sum_{\mathsf{i}=1}^\infty \mathsf{p}^\mathsf{m}_\mathsf{i}, \;\; \mathsf{p} \in 
abla_\infty$$

is loosely called the homozygosity of order **m**. The name is taken from population genetics where the homozygosity corresponds to  $\mathbf{m} = 2$ . It represents the probability that all samples are of the same type when a random sample of size **m** is selected from the population.

## Definition

The function is closely associated with the Shannon entropy in communication, the Herfindahl-Hirschmam index in economics, and the Gini-Simpson index in ecology. It provides a measure of concentration of the population in terms of individual types with large values corresponding to higher concentration.

If the proportions are random, then homozygosity becomes a random variable.

Assume that the proportions of individual types follow distribution  $PD(\theta)$ . The focus of this talk will be the random homozygosity

 $H(P(\theta); m).$ 

## LLN

# **Question**: What is the asymptotic behaviour of $H(P(\theta); m)$ when $\theta$ tends to infinity?

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# LLN: $H(P(\theta); m) \rightarrow 0$ in probability, $\theta \rightarrow \infty$ .

$$rac{ heta^{{f m}-1}}{\Gamma({f m})}{f H}({f P}( heta);{f m})
ightarrow 1$$
 in probability,  $heta
ightarrow\infty.$ 

## Gaussian Limit

#### Theorem (Joyce, Krone and Kurtz (02))

$$\sqrt{ heta}[rac{ heta^{\mathbf{m}-1}}{\Gamma(\mathbf{m})}\mathsf{H}(\mathsf{P}( heta);\mathbf{m})-1]\Rightarrow\mathsf{Z}_{\mathbf{m}}$$

where  $Z_m$  is a normal random variable with mean zero and variance

$$\frac{\Gamma(2\mathbf{m})}{\Gamma^2(\mathbf{m})}-\mathbf{m}^2.$$

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## Gaussian Limit

$$\mathsf{H}(\mathsf{P}(\theta);\mathsf{m}) \approx \frac{\mathsf{\Gamma}(\mathsf{m})}{\theta^{\mathsf{m}-1}} + \frac{\mathsf{\Gamma}(\mathsf{m})}{\theta^{\mathsf{m}-1/2}} \mathsf{Z}_{\mathsf{m}}$$

and

$$\frac{\theta^{\mathbf{m}-1}}{\Gamma(\mathbf{m})} \mathbf{H}(\mathbf{P}(\theta);\mathbf{m}) \approx \mathbf{1} + \theta^{-1/2} \mathbf{Z}_{\mathbf{m}}$$

It is natural to investigate more refined structures associated with the limits

$$H(P(\theta); m) \rightarrow 0, \ \theta \rightarrow \infty$$

and

$$\frac{\theta^{\mathbf{m}-1}}{\Gamma(\mathbf{m})} \mathbf{H}(\mathbf{P}(\theta);\mathbf{m}) \to \mathbf{1}, \ \theta \to \infty$$

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## Large Deviations From Zero

#### Theorem (Dawson and F (06))

The family  $\{\mathbf{H}(\mathbf{P}(\theta); \mathbf{m}) : \theta > 0\}$  satisfies a LDP with speed  $\theta$  and rate function

$$\mathsf{I}(\mathsf{y}) = \left\{ egin{array}{cc} \log rac{1}{1-\mathsf{y}^{1/\mathsf{m}}}, & \mathsf{y} \in [0,1] \ \infty, & \textit{else.} \end{array} 
ight.$$

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#### **Moderate Deviations**

Let  $\mathbf{a}(\theta)$  satisfy

$$\lim_{ heta
ightarrow\infty} \mathbf{a}( heta) = \infty, \lim_{ heta
ightarrow\infty} rac{\mathbf{a}( heta)}{\sqrt{ heta}} = 0,$$

and

$$\liminf_{\theta \to \infty} \frac{\mathbf{a}^{1-\epsilon}(\theta)}{\theta^{(\mathbf{m}-1)/(2\mathbf{m}-1)}} > 0$$

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for some  $\epsilon$  in  $(0, \frac{1}{2m-1})$ .

#### **Moderate Deviations**

## Theorem (Gao and F (08)) The family $\mathbf{a}(\theta) \left(\frac{\theta^{m-1}}{\Gamma(m)}\mathbf{H}(\mathbf{P}(\theta), \mathbf{m}) - 1\right)$ satisfies a LDP with speed $\frac{\mathbf{a}^2(\theta)}{\theta}$ and rate function $\frac{\mathbf{x}^2}{2(\Gamma(2\mathbf{m})/\Gamma(\mathbf{m})^2 - \mathbf{m}^2)}$ , $\mathbf{x} \in \mathbf{R}$ .

#### Remark

Let  $\mathbf{a}(\theta) = \theta^{\delta}$ . Then moderate deviation holds for

$$heta^{\delta}\left(rac{ heta^{\mathbf{m}-1}}{\mathsf{\Gamma}(\mathbf{m})}\mathsf{H}(\mathsf{P}( heta),\mathbf{m})-1
ight)$$

if and only if  $\delta \in (\frac{m-1}{2m-1}, \frac{1}{2})$ .

This indicates a significant departure from the Gaussian regime when  $\delta$  is between 0 and  $\frac{m-1}{2m-1}$ .

## Large Deviations From One

The case  $\delta = 0$  corresponds to the large deviations of

$$rac{ heta^{\mathsf{m}-1}}{\Gamma(\mathsf{m})}\mathsf{H}(\mathsf{P}( heta),\mathsf{m})$$

from one.

#### Fundamental Differences From LDP for $H(P(\theta), m)$

- The state space is no longer compact
- Exponential tightness is not free
- Do not have exponential moment in the neighbourhood of zero

## Large Deviations From One

#### Theorem (Dawson and F(16))

A large deviation principle holds for  $\frac{\theta^{m-1}}{\Gamma(m)}H(P(\theta);m)$  as  $\theta$  converges to infinity on space R with speed  $\theta^{1/m}$  and good rate function

$$\mathbf{S}(\mathbf{x}) = \left\{ egin{array}{ll} [\Gamma(\mathbf{m})(\mathbf{x}-1)]^{1/\mathbf{m}}, & \mathbf{x} \geq 1, \ +\infty, & ext{otherwise}. \end{array} 
ight.$$

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Note: The scale of deviations for  $\mathbf{x} < 1$  is different from that of  $\mathbf{x} > 1$ .

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## Link Between the Two Rate Functions

**Question:** Can one derive the LDP for  $\frac{\theta^{m-1}}{\Gamma(m)}$ **H**(**P**( $\theta$ ); **m**) from the LDP for **H**(**P**( $\theta$ ); **m**) or vice versa?

Answer: ??

## Link Between the Two Rate Functions

Recall that the LDP for  $H(P(\theta); m)$  has speed  $\theta$  and rate function

$$\mathbf{I}(\mathbf{y}) = \left\{ egin{array}{cc} -\log(1-\mathbf{y}^{1/m}), & \mathbf{y} \in [0,1] \\ \infty, & ext{otherwise} \end{array} 
ight.$$

Since  $\mathbf{H}(\mathbf{P}(\theta); \mathbf{m})$  and  $\mathbf{H}(\mathbf{P}(\theta); \mathbf{m}) - \frac{\Gamma(\mathbf{m})}{\theta \mathbf{m}-1}$  are exponentially equivalent, the same LDP holds for  $\mathbf{H}(\mathbf{P}(\theta); \mathbf{m}) - \frac{\Gamma(\mathbf{m})}{\theta \mathbf{m}-1}$ .

#### Link Between the Two Rate Functions

Write  $\frac{\theta^{m-1}}{\Gamma(m)} \mathbf{H}(\mathbf{P}(\theta); \mathbf{m})$  as

$$\frac{\theta^{\mathsf{m}-1}}{\Gamma(\mathsf{m})}[\mathsf{H}(\mathsf{P}(\theta);\mathsf{m})-\frac{\Gamma(\mathsf{m})}{\theta^{\mathsf{m}-1}}]+1.$$

For  $\mathbf{x} \in [1,\infty)$  and  $\frac{\theta^{m-1}}{\Gamma(m)}\mathbf{H}(\mathbf{P}(\theta);\mathbf{m}) = \mathbf{x}$ , let  $\mathbf{y} = \frac{\Gamma(\mathbf{m})}{\theta^{m-1}}(\mathbf{x}-1)$ . Then

$$\begin{split} \exp\{-\theta \mathbf{I}(\mathbf{y})\} &= & \exp\{-\theta^{1/\mathbf{m}+\mathbf{m}/(\mathbf{m}-1)}\log\frac{1}{1-(\frac{\Gamma(\mathbf{m})}{\theta^{\mathbf{m}-1}}(\mathbf{x}-1))^{1/\mathbf{m}}}\}\\ &\approx & \exp\{-\theta^{1/\mathbf{m}}\mathbf{S}(\mathbf{x})\}. \end{split}$$

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## Main Steps of Proof

#### Step 1

Showing that LDP for general  $\theta$  is equivalent to  $\theta$  being integers.

#### Step 2

For integer  $\theta$ , find a new representation of  $\frac{\theta^{m-1}}{\Gamma(m)} H(P(\theta); m)$  as

$$\frac{\theta^{m-1}}{\Gamma(m)} \mathbf{H}(\mathbf{P}(\theta); \mathbf{m}) = \frac{\theta^{m-1}}{\Gamma(m)} [\frac{1}{\gamma^{m}(\theta)} \sum_{\mathbf{k}=1}^{\theta} \mathbf{W}_{\mathbf{k}}^{m} \mathbf{H}_{\mathbf{k}}]$$

where  $\mathbf{W}_1, \ldots, \mathbf{W}_{\theta}$  are independent copies of  $\gamma(1)$ , and independently,  $\mathbf{H}_1, \ldots, \mathbf{H}_{\theta}$  are independent copies of  $\mathbf{H}(\mathbf{P}(1); \mathbf{m})$ .

## Main Steps of Proof

#### Step 3

Exploring the independence and the LDP for gamma distribution to verify that the LDP for  $\frac{\theta^{m-1}}{\Gamma(m)} \mathbf{H}(\mathbf{P}(\theta); \mathbf{m})$  is equivalent to the LDP for

$$\frac{1}{\Gamma(\mathbf{m})\theta}\sum_{\mathbf{k}=1}^{\theta}\mathbf{W}_{\mathbf{k}}^{\mathbf{m}}\mathbf{H}_{\mathbf{k}}$$

Step 4

Applying Cramér's theorem for  $\mathbf{x} < 1$ .

Step 5

Applying Nagaev's result for  $\mathbf{x} > 1$ .

## Generalizations

#### What about other random distributions?

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