



Об участии в  
European Conference of  
Human Genetics 2015

6-9 мая, Глазго, Великобритания

Георгий Романов, асп. 1 года,  
НИЛ Молекулярной биологии, ИЕН, СВФУ



- В этом году конференция проходила в Глазго, Шотландия.





Местом конференции был  
Scottish Exhibition and Conference Center



Конференция впечатляет масштабами,  
2700 участников  
1454 постерных доклада  
145 организаций на выставке.

# Наши постеры

**02.49**

**Identification of novel mutation p.W325X (c.977G>A) in the POU3F4 gene with perilymphatic Cochlear-deafness syndrome (DS-32) in Yakut family (Eastern Siberia)**

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**Introduction**

The most rare causes of deafness in the world are X-linked and mitochondrial forms of hearing impairment (1-2%). For the first time, we revealed identical abnormalities of inner ear in two brothers from Yakut family (Eastern Siberia) with presumably X-linked recessive deafness that are typical for the perilymphatic Cochlear-deafness syndrome (DSNX2, OMIM304400) (Fig. 1).

**Materials and methods**

**Results and Discussion**

Computed tomography studies demonstrated an abnormal dilatation of the internal acoustic canal (IAC) as well as an abnormally wide communication between the IAC and the inner ear compartment in both brothers (Fig. 2A). Parents had no any temporal bone anomalies. To determine whether mutations in the POU3F4 gene (Xq21.1) are responsible for the hearing loss in these patients, we sequenced the single exon of the POU3F4 gene. We identified novel nucleotide substitution c.977G>A in POU3F4 in heterozygous state in both probands, in the heterozygous state in their mother, and c.977G>A was absent in their father (Fig. 2B). Transition c.977G>A leads to stop-codon (p.W325X) in the POU3-homeodomain of human transcription factor protein (Brain 4) (Fig. 3,4).

**Conclusions**

Segregation of inner ear malformations and novel mutation c.977G>A (p.W325X) in the POU3F4 gene with deafness in studied Yakut family confirmed association of the POU3F4 gene with clinical phenotype of perilymphatic Cochlear-deafness syndrome (DSNX2, OMIM 304400).

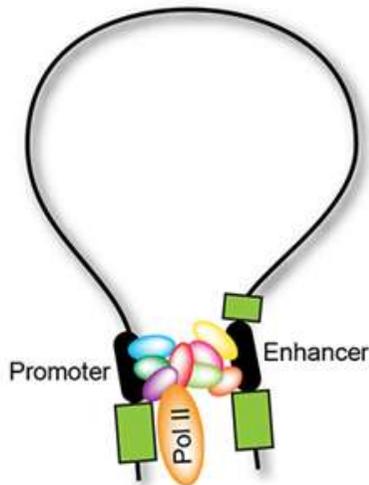
**ACKNOWLEDGEMENTS**

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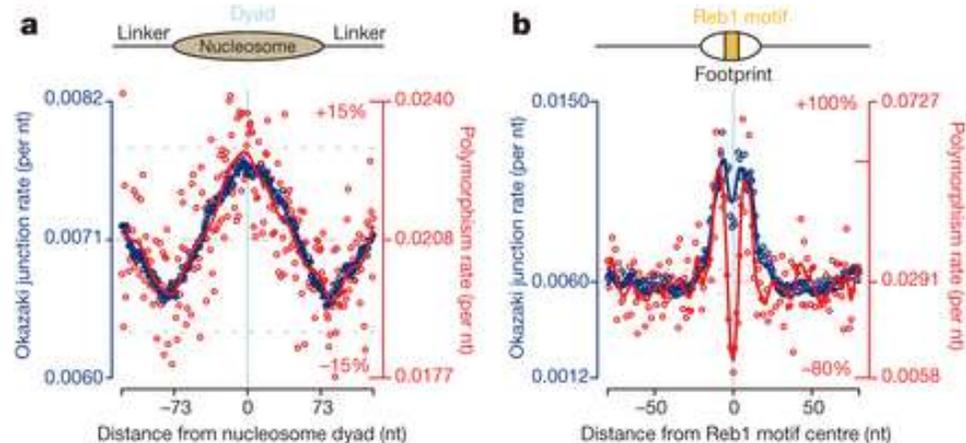
## Интересные доклады



- **Chromosome conformation and long-distance gene regulation**
- N. Benabdallah, S. Bhatia, I. Williamson, **W. Bickmore**;
- О регуляции генов энхансерами находящимися на расстоянии до 1 мб, которые могут находиться в межгенной или интронной области. И нарушения в этих отдалённых областях приводят к дисфункции целевого промотора. Взаимодействие же между ними регулируется разными вариантами укладки самой ДНК с белками в хромосоме и в ядре.



## Интересные доклады



- **Ribonucleotides embedded in genomic DNA**
- **A. Jackson**
- При каждой репликации цепочки геномной ДНК, в последовательность случайно встраивается около миллиона РНК. Такие рибонуклеотиды являются неканоничными и удаляются при помощи белкового комплекса, кодируемого тремя генами RNASE H2. Эти же рибонуклеотиды являются важными точками хромосомной нестабильности. Авторы исследования разработали новую технику которая позволяет использовать такие внедрённые рибонуклеотиды, чтобы отслеживать соответствующий вклад ДНК полимеразы в уровень локальных замен нуклеотидов в функционально важные регионы, влияющий на мутационный ландшафт генома.

# Интересные доклады

- The role of telomeres in aging
- C. Bar;
- Исследование проведено на модели действия TERT на трансгенных мышей. В первом варианте мыши сами содержали высокоактивную TERT, и в совокупности с более активными генами ингибирования опухолей. Во втором варианте использовались вирусные векторы AAV с внедрённой TERT, действию которых подвергали мышей. В итоге были получены мыши живущие на 40% дольше.



# Одной строкой

- Ретротранспозоны LINE-1 являются одной из главных сил влияющих на эволюцию генома человека. **J. Garcia-Pérez**
- Регуляторы активности бурой жировой ткани имеют важную роль старении и развитии нейродегенеративных заболеваний. **J. W. Kornfeld**
- Модным стало применение системы CRISPR/Cas9
- Широко обсуждается проект 100 000 геномов
- Также было много других интересных докладов

Кроме научной программы было много интересных знакомств и общения



A.F. Roux



Feliciano Ramos

- Спасибо за внимание!

