

# **EXPRESSION PROFILE AND TRANSCRIPTION FACTOR BINDING SITE EXPLORATION OF GENES IMPRINTED IN HUMAN AND MOUSE**

BMC Genomics  
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## **BACKGROUND**

### **IMPRINTED GENES:**

- Represent a minority of genes, which are transcribed from only one allele;
- Play a role in the regulation of embryonic growth;
- Control placental function;
- Modulate the transport of nutrients from mother to embryo;
- Control neurological development.

## **BACKGROUND**

- Based on recent predictions, the number of mammalian imprinted genes may range between 100-600 genes.
- Imprinted genes are tightly regulated in a developmental and tissue specific manner
- The imprinted expression of genes appears to be a rather conserved phenomenon in mammals.
- Orthologous genes that are imprinted in human and mouse are most likely either maternally or paternally expressed.

## **BACKGROUND**

Computational expression analysis of human and mouse imprinted genes to:

- Compared the profiles of individual genes across tissues;
- Analysed the correlations of expression patterns in human and mouse;
- Explored the role of predicted transcription factor binding sites

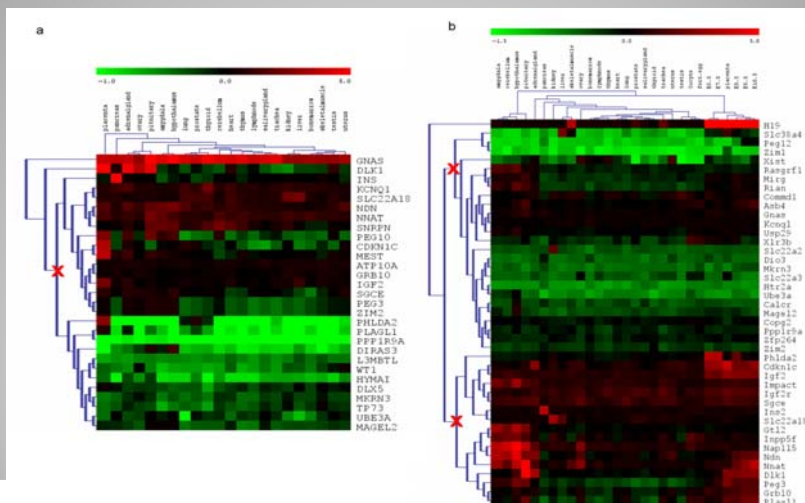
# RESULTS

## GENES SELECTED FOR ANALYSIS

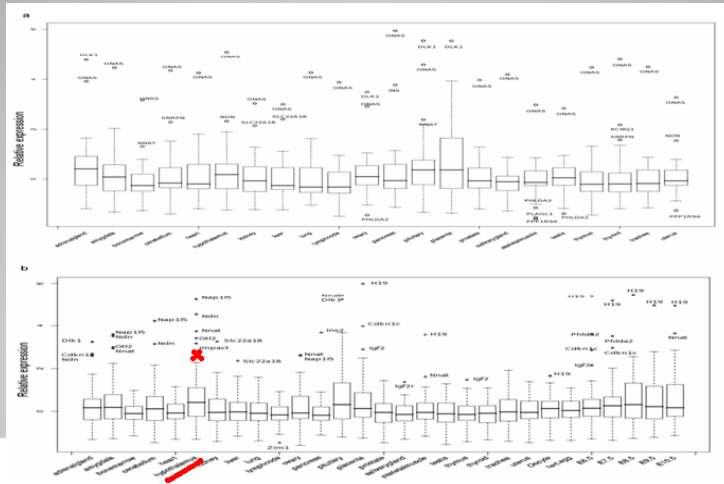
Information gathered from the IGC on 62 genes:

- For 30 genes imprinted status analysed in human and mouse → 26 imprinted in both species, 1 confirmed only in human and 3 only in mouse.
- For 23 genes imprinted status analysed in mouse.
- For 9 genes imprinted status analysed in human.

## TISSUE-SPECIFIC EXPRESSION PATTERNS OF IMPRINTED GENES

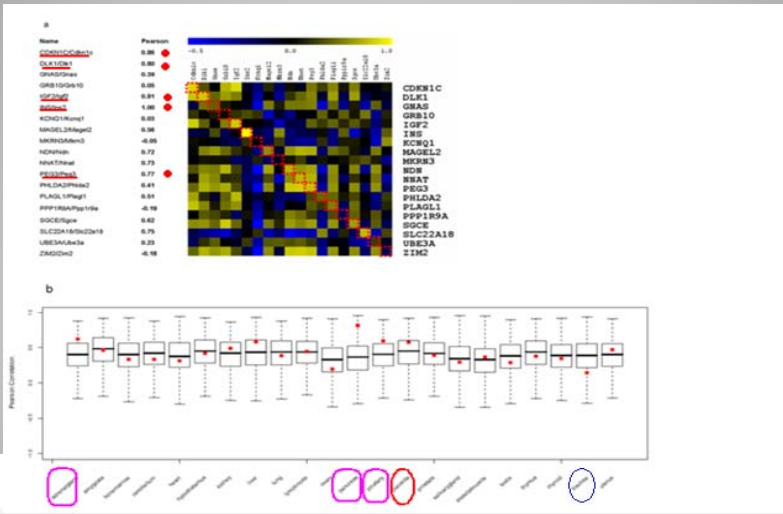


## IMPRINTED GENES DO NOT SHOW PROMINENT OVEREXPRESSION IN DISTINCT TISSUES

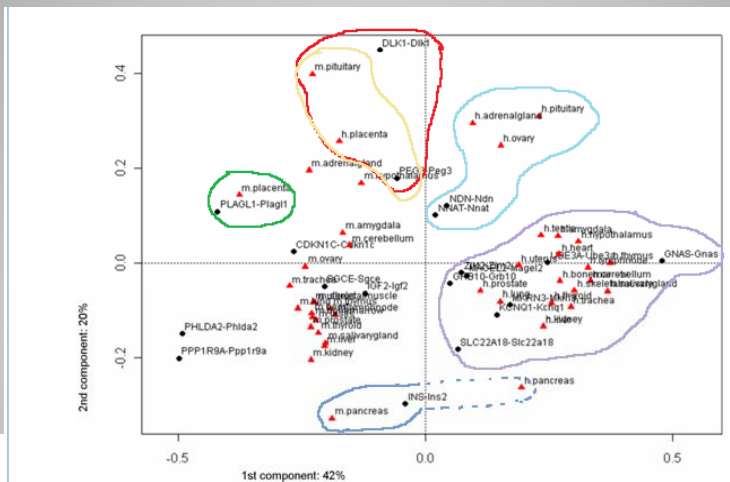


- Maternally and paternally imprinted genes do not cluster together.
- The parental origin of expression has no influence on tissue-specific expression profile.
- Paternally expressed genes tend to be more highly expressed than maternal genes.

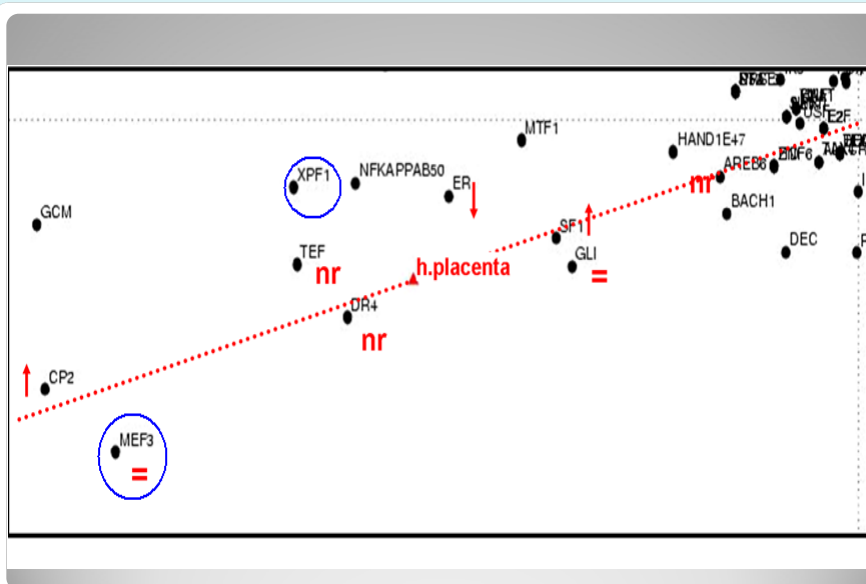
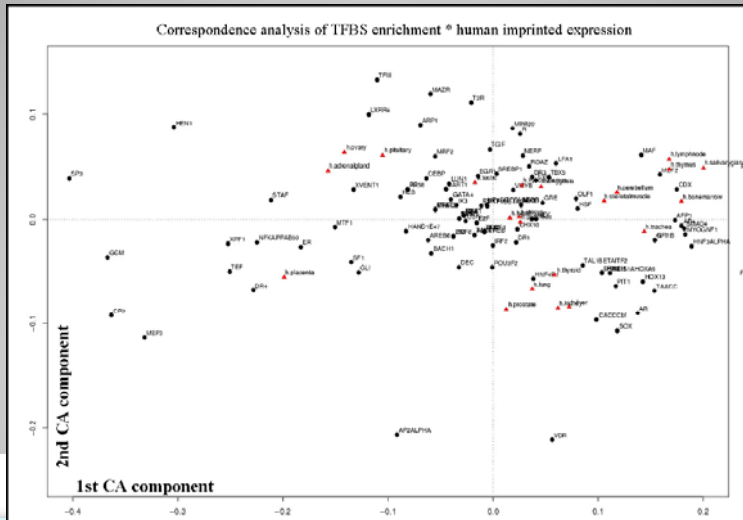
# CORRELATION OF ORTHOLOGOUS GENE EXPRESSION IN HUMAN AND MOUSE

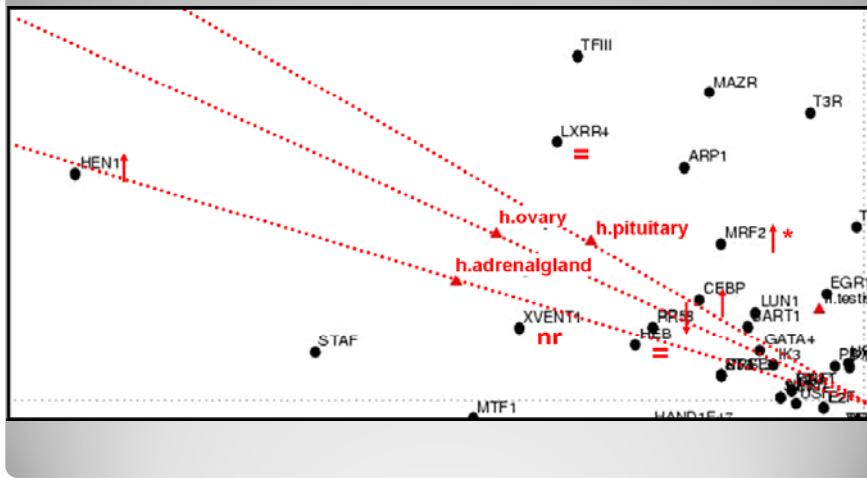


# CORRESPONDENCE ANALYSIS OF RELATIVE GENE EXPRESSION OF ORTHOLOGOUS IMPRINTED GENES



## CORRESPONDENCE ANALYSIS OF RELATIVE EXPRESSION AND TFBSs OF HUMAN ORTHOLOGOUS IMPRINTED GENES





## DISCUSSION

- ❖ Imprinted genes are expressed in a broad range of adult tissues and placenta in human and mouse.
- ❖ Imprinted genes orthologues do not show a strong correlation of tissue-specific expression patterns in human and mouse.
- ❖ The correlation of expression patterns of imprinted genes in human and mouse is not pronounced and does not differ from that of randomly genes.

## DISCUSSION

- ❖ Tissues with distinct expression profile are pituitary, adrenal gland, pancreas, placenta and ovary.
- ❖ Imprinted genes are strongly expressed in the pituitary, adrenal gland, pancreas and placenta.



Role in the energy metabolism and homeostasis of mammals

## DISCUSSION

- ❖ Pituitary, adrenal gland, placenta and ovary show a remarkable correlative association with distinct TFBSs in the promoter region of imprinted genes.