

174
N 28/rg

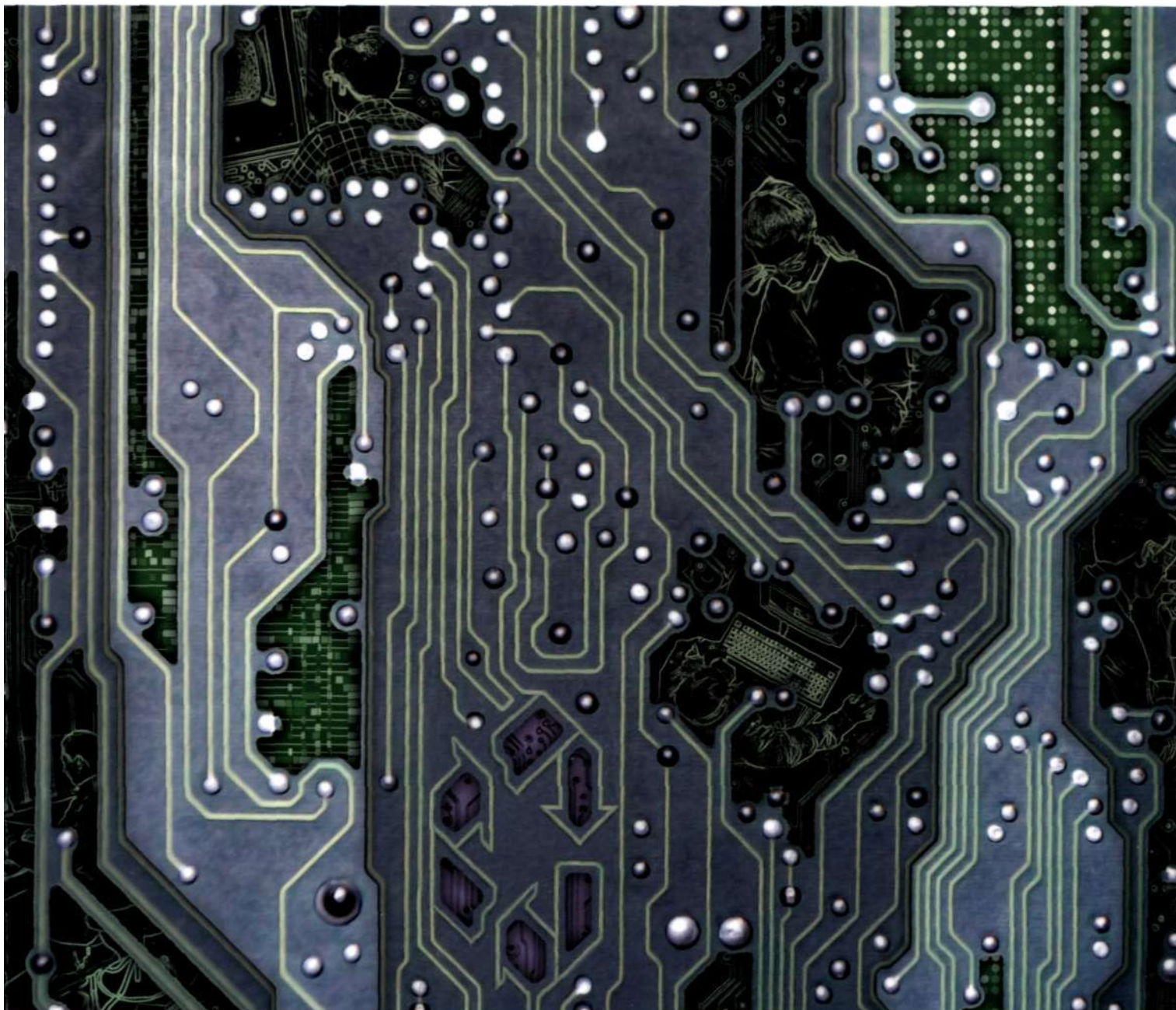
nature

REVIEWS

february 2013 volume 14 no. 2

www.nature.com/reviews

GENETICS



**REUSING GENE
EXPRESSION DATA**

Opportunities and pitfalls

**Genomic structural
variation**

Implications for human health



The thorny issue of heritability in human disease p139

CONTENTS

February 2013
volume 14 no. 2

REVIEWS

89 Reuse of public genome-wide gene expression data

Johan Rung and Alvis Brazma

A wealth of microarray gene expression data and a growing volume of RNA sequencing data are now available in public databases. The authors look at how these data are being used and discuss considerations for how such data should be analysed and deposited and how data reuse could be improved.

125 Phenotypic impact of genomic structural variation: insights from and for human disease

Joachim Weischenfeldt, Orsolya Symmons, François Spitz and Jan O. Korbel

With the increased cataloguing of human structural variants, our understanding of their influence on phenotype is ever improving. Here, the influence of structural variants on phenotypes including disease is discussed, and strategies for further characterization are presented.

100
FEATURED
ARTICLE

RNA interference in the nucleus: roles for small RNAs in transcription, epigenetics and beyond

Stephane E. Castel and Robert A. Martienssen

In addition to well-known roles in the cytoplasm, a growing number of functions for small RNAs in the nucleus are being discovered. These include roles in transcriptional repression, epigenetic modifications and genome stability. This Review considers examples from animals, plants and fungi.

139 The heritability of human disease: estimation, uses and abuses

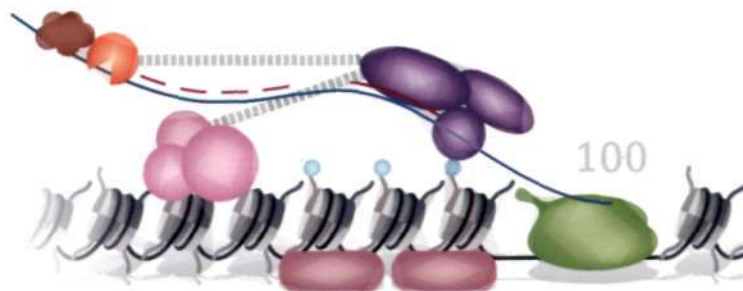
Albert Tenesa and Chris S. Haley

Heritability estimates provide a useful means of understanding the genetic and environmental contributions to phenotypic variance. The authors define heritability, discuss how to estimate and interpret it in the context of disease and examine how biases in heritability estimates arise.

113 Y-chromosome evolution: emerging insights into processes of Y-chromosome degeneration

Doris Bachtrog

The Y chromosomes of many species, including humans, are gene-poor and degenerate. The recent application of genome-wide technologies to evolutionarily old and young Y chromosomes has provided insight into the processes that have shaped them and their future.



On the web www.nature.com/reviews/genetics

Advance online publication

We operate an advance online publication (AOP) service for authors and readers to view the latest articles published online ahead of print.

Forthcoming articles:

Sequence assembly demystified

Niranjana Nagarajan and Mihai Pop

Genotype to phenotype: lessons from model organisms for human genetics

Ben Lehner

DNA methylation: roles in mammalian development

Zachary D. Smith and Alexander Meissner

Nature Reviews Genetics (ISSN 1471-0056, USPS 019269), is published monthly by Nature Publishing Group, Porters South, 4 Crinan Street, London N1 9XW. The 2013 US annual subscription price is \$4677 (Full), \$265 (Personal 1 year) and \$199 (Student). Airfreight and mailing in the USA by agent named Air Business Ltd, c/o Worldnet Shipping Inc., 156-15, 146th Avenue, 2nd Floor, Jamaica, NY 11434, USA. Periodicals postage paid at Jamaica NY 11431. US Postmaster: Send address changes to Nature Reviews Genetics, Air Business Ltd, c/o Worldnet Shipping Inc., 156-15, 146th Avenue, 2nd Floor, Jamaica, NY 11434, USA. Subscription records are maintained at Nature Publishing Group, Brunel Road, Basingstoke, Hampshire RG21 6XT, UK. Air Business Ltd is acting as our mailing agent.

SERIES ON NON-CODING RNA

<http://www.nature.com/nrg/series/noncoding>

SERIES ON STUDY DESIGNS

<http://www.nature.com/nrg/series/studydesigns>

Links to further information

The full text of articles includes author biographies, links to glossary terms and links to websites and databases with relevant information.

Twitter

Follow *Nature Reviews Genetics* on Twitter for the latest news from the journal and beyond: @NatureRevGenet

Online correspondence

Comments from readers about our articles. All correspondence will be highlighted on the table of contents and on e-mail alerts.

RESEARCH HIGHLIGHTS

77 Selections from the recent scientific literature

PROGRESS

83 The evolutionary causes and consequences of sex-biased gene expression

John Parsch and Hans Ellegren

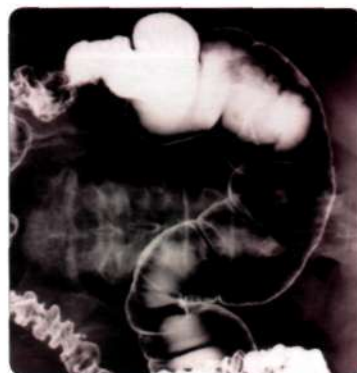
The authors discuss recent advances — mainly gained through genomic methods such as RNA sequencing — in our understanding of the causes and consequences of differences in gene expression between females and males. Areas of progress include our understanding of the roles of sexual antagonism and dosage compensation.



Getty



COVER: 'Data-based' by Patrick Morgan, inspired by the Review on p89.



Photodisc/Getty

Microbiome variants get personal p80

EDITORIAL OFFICE

LONDON: NatureReviews@nature.com
The Macmillan Building, 4 Crinan Street,
London N1 9XW, UK
Tel: +44 (0)20 7843 3620;
Fax: +44 (0)20 7843 3629

To subscribe and for more detailed information visit
www.nature.com/reviews/genetics

CHIEF EDITOR: Louisa Flintoft
SENIOR EDITOR: Mary Muers
ASSOCIATE EDITOR: Hannah Stower
ASSISTANT EDITOR: Darren Burgess
SENIOR COPY EDITOR (NRG): Matthew Smyllie
SENIOR COPY EDITORS: Mariam Faruqi,
Cattriona Rodwell, Isabel Woodman, Lucie Wootton
COPY EDITING MANAGER: Lewis Packwood
SENIOR ART EDITOR (NRG): Patrick Morgan
SENIOR ART EDITORS: Vicky Summersby, Kirsten Lee
ART CONTROLLER: Susanne Harris
MANAGING PRODUCTION EDITOR:
Judith Shadwell

SENIOR PRODUCTION EDITOR: Simon Fenwick
PRODUCTION CONTROLLER: Natalie Smith
OFFICE MANAGER: Laura Lee
MANAGING EDITOR: Suzanne Farley
SENIOR EDITORIAL ASSISTANTS: Laura Corns,
Ella Lines
WEB PRODUCTION MANAGER: Dipti Shah
MARKETING MANAGERS: Tim Redding,
Virginia Lee
PUBLISHER: James Butcher
CUSTOMER SERVICES: Feedback@nature.com

Copyright © 2013 Nature Publishing Group
Printed in Wales by Cambrian Printers
on acid-free paper.

EDITORS



LOUISA FLINTOFT



MARY MUERS



HANNAH STOWER



DARREN BURGESS