

)LQDO 5HSRUW

8WLOLJLQJ DQG 7HDFKLQJFORGOL\$OU1K1Q8PUSV IRU (QYLU

6WUHV 7ROHUDQFH

)HOL[ )ULWVFKL 8QLYHUVLW\ RI 0LVVRXUL  
+XVVHLQ 6KLPVOLV 8QLYHUVLW\ RI .ZD=XOX

6<1236,6

7KH PDMRULW\ RI SODQW EUHHGHUV DQG SODQW VFLHQWL  
&HQWHU IRU &URS ,PSURYHPHQW \$&&, ORFDWBG DW=WKH  
LQ 6RXWK \$IULFD \$&&, WUDLQV SRVWJUDGXDWH VWXGHQV

IRXQGDWLRQ

IRU IXWXUH FROODERUDXMRLO 6ZHBK1D1W1V1W1D1Q1S  
6FLH1D1G 'HSXW\ \$IULFD1Q1 &HQW,PSURYH1D1Q1SVKHU81Q1W RI  
.ZD=X1D1W1D1O

1DWD0 DQG

KDG D GHVLUH WR YLVLWRWKH WRK1D1G1D1W1V1D1Q1G1V

VKDGRZVHGH ODE P1D1E1W1U1Q1X1F1W1H1G1K1L1R1K1J1S1X1W  
SK1R1W1\1S1D1D1D1Q1P1H1G1H1Q1W1L1G1L1Q1J1P1R1W1R1G1H1R1D1X1Q1W1W1R1\1E1H1D1Q1J1H1C  
,Q1D1G1G1L1W1L1R1Q1W1R1O1H1D1U1Q1L1Q1D1W1D1R1X1W1S1K1B1C1P1H1M1S1R1E1U1S1K1L1P1H1O  
H1Q1J1D1G1Z1L1W1K1'1U1.1U1L1V1D1Q1L1G1K1B1D1W1H1K1D1U1Q1D1E1R1X1W1K1W1D1D1W1H1P1H1W1K1R1G1Y  
V1R1\1E1H1D1Q1O1H1F1X1O1D1U1E1L1R1O1R1J1U1P1D1Q1J1H1G1E1B1E1R1L1R1U1D1Q1S1R1L1W1H1V1H1D1U1F1K1  
31U1R1I1H1V1R1U1V161K1D1E1Q1G1L1Q1W1K1H1K1R1U1W1N1E1R1D1E1R1H1U1Q1\$1X1E1D1E1E1H1H1G1L1Q1J  
S1U1R1J1U1D1P1H1V1H1L1Q1W1S1W1D1R1F1W1G1R1G1V1X1V1H1I1R1O1L1Q1V161K1B1W1H1O1H1D1U1F1K1D1Q1C  
W1H1D1F1K1L1Q1J1H1I1R1U1W1Q1G1H1L1Q1V1O1X1G1\$1D1Q1W1L1R1J1H1G1J1H1D1S1S1J1R1O1F1K1H1F1V  
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3URIRU V6KLPVOLV DQG 'U )ULWVFKL KDYHUEH1K1Q1P1H1O1U1W1J1X1O1D  
U1H1W1X1U1Q1H1G1W1H1D161R1R1V1Q1W1\$1Q1X1H1W1R1L1G1H1Q1W1L1I1\1R1S1S1R1I1U1D1W1K1Q1Q1V1L1H1V  
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ORZ RUOXRQWUDDILQRVH IDPLO\ RI ROLJRVDFFKDULGHV 7KH  
DVVLVWHG VHGHSFWRVPHDQGR I QHZ VR\EHDQ RU UHODWHG F  
IHHG SURGXFWLRQ

, DOVR KDG WKH RSSRUWXQLW\ WR OHDUQ IURP 'U %LO\HX  
FURVVLQJ EQRIKWURDQWQGXFHG PXVEDQHWGLDQSRISKEDSVLHR  
JHQRW\SH VDPSOLQJ IRU PROHFXODU JHQHWLF DQDO\VLV  
)XUWKHUPRUH , DWWHQGHG WKH ZHHNO\ ELRLQIRUPDWLF\  
RQ WKH SURJUHVV DQG RXWFRPHV RI WKELGHQDQVIRVLDQWLF  
VWXRG\ 'UV %LO\HX\ V DQG 6FDERR\ V JURXS DQG WKHLU 3K' F

'U %LO\HX\ V ZRUN RQ JHQH GLVFRYHULHV PDLQWHQDQFH D  
UHVXRUFHV JHQHWLF DGYDQFHPHQW DQG SUHFLVLRQ EU  
GHYHORSPHQWLPISQRZHQGURS FXOWLYDUV , WUHDVXUH L  
PDNH RXU UHVHDUFK SURJUDPV LQQRYDWLYH DQG DW WK  
VFLHQFH

Genomic data analysis

, GUDIWHG D WH[WERRN RQ UHVHDUFK PHWKRGRORJ\ IRU W  
LQ DJULFXOWXUDO VFLHQFHV SRVWJUDGXDWH VWXGHQW  
DJULFXOWXUDO HGXFDWRUV DQG UHVHDUFKWLUVHQLJEFDQGH  
DQG JHQRPLF GDWD DQDO\VLV GXULQJ WKLV VFLHQWLILF  
UHYLHZHEGWLKHSULQRIOSHFXDURU JHQHWLF SDUDPHWRU DQG  
PROHFXODU GDWD DVVHPEO\ DQG DQDO\VLV WRLFRVFXWDHQG  
GHGXFH SRSXODWLRQ VWUXFWXUHV RI WHVW JHQRW\SHV L  
GDWD DQDO\VLV ZHUH FUXFLDO WR GHYHORSLQJ WKH GUD  
XQGHUJUDGXDWH DQG SRVWJUDGXDWH SODQWD EDQDQV





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B

C

D

)LJXUHR\EHDQ URRW SKHQRW\SLQJ DW 5ROOLQV ERWWRP  
\$ URRW LPDJH DQDO\VLV IURP GLIIHUHQW DQJOHV GURC  
,

1HWZRUNLQJ DQG FRSSORDJ\RXU\DLWLRQ\

x ,Q WKH ODVW ZHDNLRIY\XO\UG\WRID\H 0HUVKD\TV ODE DW W  
RI &LQFLQQDWL 2KLR JH\OH\DD\QHGQW\SHD\XHHQ\WRQQIRUPDWL  
WRROV DQG FORXG FRPSXWLQJ SU\WJ\DDPWO\XWH\RXOD OR UHEV  
SODQW JH\Q\YR\OLFLVQJ PXOWLGLVFLSOLQDU\ WHDPV

x , YLVLWHG WKH 8QLYHUVI&K\DRSD\DJQLQ8\B& 8P&D&XVW  
DOORZHG PH WR GLVFXVV ZLWK W&K\HUGRAE\BIDK, Q&Q&O&D&R&E  
DQ&HH W&B& FDPSXV DQG 5HVH&B&D&SKUR&G&K&FLWEXMVLQR ZLWK  
6,/ WHDP LQF&X&H&L&H&U \*R&O&GVPLWK %ULDQ 7KHUDE&B&Q&G 5LV  
YLVD&G LQWH&ZLD&F&W&SR&W&H&QWLD&OO\ IRUJH FROODERUDWLRQ

7KURXJK W&K&H&D&G&O&H&R&W&H FURSSLQJ V\VVWHPV VSDQQQLQJ WK  
DQG ,OOL&Z&R&V\H[FLWHG&H&Z&R&J&V&G&H&U&D&W&H&L&R&Q PDLJH YDULHW\ Z  
SODQW HUHFW&H&O&B&D&L&W&H&S&H&VKR&D&U&G&S&O&P&H&Q&W&K&E&H&J&K&D&Q&W  
GHQV\W&X&U&H



x 'U )HOL[ )ULW ~~WFKD~~FDVHG 0HJKQDWK IQDQFDQ\$QBUHZ 6DFEF  
DQG WKH %D\ )DUP WHDP &BUQOVREQ DQGHIDVOS  
x 'U 5LWD 0IXRPUUDQJLQJ DR\GYK ~~WRW~~&QJ  
x 'U 7HVID\H 0HRUV ~~WX~~HDQGYK ~~RHW~~KGJ8QRYHUFLLQVDRM &LQ  
x 7KH VWDII RI WKH 8QLYHUVLW\ \$LU%Q% 1DQFS®%N06WD