

# 85FOBUBINTDSFFOJOH BOE EJBHOPTUID UFTUIOH OPS OF UBM DI SPN PTPN BINBOE HFOFUID DPOEJUPOT

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- %2##2&;ZR[^Y MUUZ\_T[a)PNQQYY aZUOMOPa\_UZSOOM #\_UY \XQNZPQ[Z\_U\_OZ`XNZSaNSOCTOZ PU\_Oa\_UZSUZbQ\_USMUUZ\_t c UT Q[ZRYY MUUZ`TM\_`TQUZR[^Y MUUZ TM\_NOOZ aZPQ^`[[PNZP c UT `TM aZPO^\_NZPUZS P[OaY OZ`OPž
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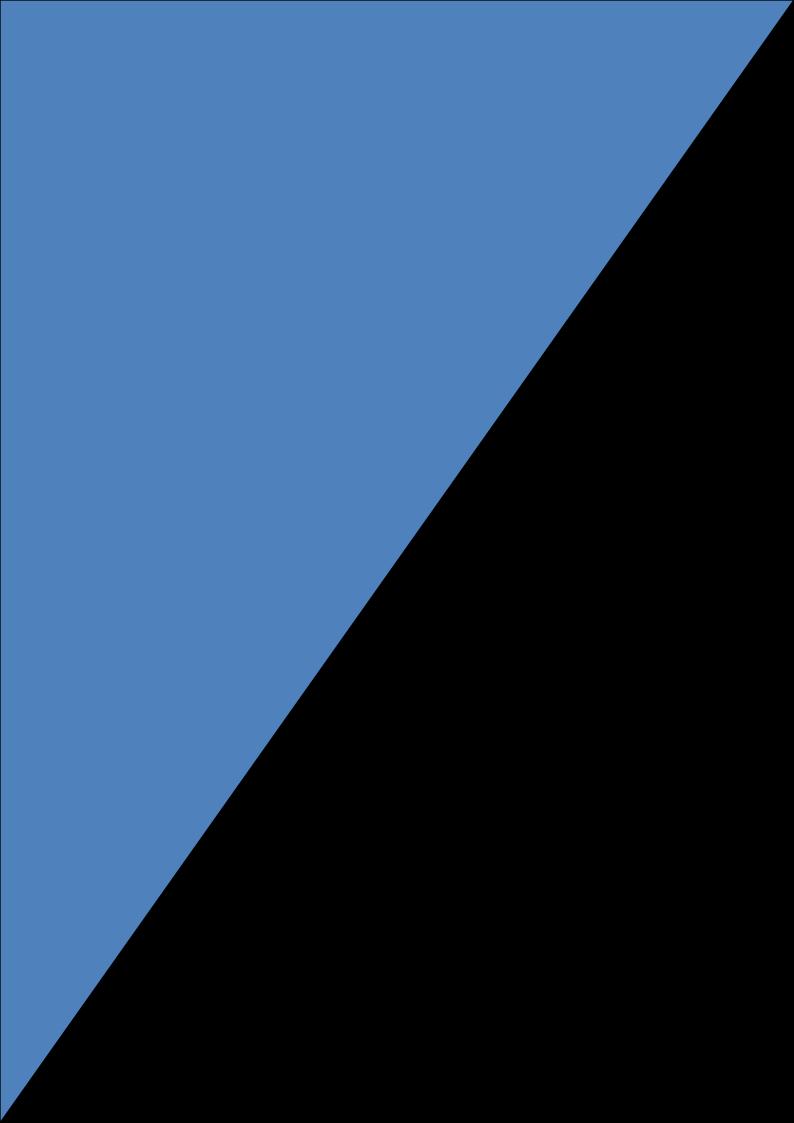
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#### 3.5.1 Population-based reproductive carrier screening

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 $\begin{array}{l} B^{\prime}QQ[ZQQ_{\ U}Z[^{\ 0}^{\ 0}ZMNXSQZQ_{\ U}Q_{\ 0}^{\ 0}QQZ_{\ U}ZS[RQ[a_{\ Q_{\ U}}CWXUPQZ_{\ U}Re^{T[_Qc_{\ U}T_{\ MZ}}] \\ RSUBUZS_{\ NU^{\ T}}[ MOTUR_{\ C}UT_{\ M}^{\ 0}QQRDTQ^{\ U}MXQPU[^{\ 0}PQZ_{\ C}^{\ C}PQZ_{\ C}^$ 

 $\label{eq:local_solution_constraint} RNSUQJ_eZP^{Y} Q_{Q} Q_{D} Q_{D}$ 

[ZQ^000\_Ub0PU[^P072<sup>55</sup>

$$\label{eq:linear_cond} \begin{split} & UT `TQUZ` ^[PaOU]Z [RZQ: SQZ[Y ID_Q] a QZQUZS `QQTZU] a Q_{1} QM^{1}Q^{2}QQQZUZS R ^{1} ^MY a XU a PQ [R \\ ^QQQ_ IbQQ[ZPUU]Z_UZ[c NbN0KNNQR ^{1} ^Q[a XQ_c T[TNbQZ[RWTU & TU] ^{e} [RNSQZQ ID PU] ^{PQ2} ^{2\#} FTU \\ _[ZQN%QP cQd NZ POP QM^{1}Q^{2}QQZUZSp_T[a XP [Z & NQ[RQ2 OP UZ `TQQ[Z`Qd` [Rc Q%ZPQRZ OP OKZUDNK \\ ^MTc Ne_R ^{1} ^{O} ^{QZ}NZP ^{[Z}Q_ SQZQ ID Q[ a Z_Q%ZS M_a `[ $& [RNPa X_c UK Q_ `[ UIbQR ^{1} ^M XQM] \\ ~[ZQA COD UIC OP UI (D C C ^{4}) \\ ~[ZQA COD UIC OP UI (D C C ^{4}) \\ ~[ZQA COD UIC OP UI (D C C ^{4}) \\ ~[ZQA COD UIC OP UI (D C C ^{4}) \\ ~[ZQA COD UIC OP UIC OP C ^{4}) \\ ~[ZQA COD UIC OP UIC OP C ^{4}) \\ ~[ZQA COD UIC OP UIC OP C ^{4}) \\ ~[ZQA COD UIC OP C ^{4}) \\ ~[ZQA$$

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#### 4. Governance

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:  $M_0 a \tilde{Z} [Z_PMQVZ] c XQPSQMN[a^TQQa^QZ^_OQZVZSY [PNXUUQ_N_0MXMVQNZPVZ c TM] _Q^UZ_S_TQe QNZ NQVY NXQY QZ^QPZ$ 

 $\label{eq:linear} 4QMNXQ^{[\[\]}bUPQ^{Q}XZP^{[\]}ZQ^{U}XP^{V}MUZI_a^{[\]}MZPQ[aZ_QXXZS]UZQAPUZSc^{U}QZ^{Q}APUZSC^{U}QZ^{Q}APUZSC^{U}QZ^{Q}AP$ 

 $\begin{aligned} & \mathsf{BM}^{\mathsf{L}}(\mathsf{D}) \mathsf{M}^{\mathsf{L}}(\mathsf{Z}}(\mathsf{Z}^{\mathsf{L}}(\mathsf{Z}^{\mathsf{L}}(\mathsf{Z}^{\mathsf{L}}$ 

 $: OVXT \land [RQ_UZNX_ \land [bUPUISOMQ`[ \land OSZNII` c [Y OZ c UXKNOZOPU R'[Y aZPQ^ MUIUS_[Y QY [PaXQ_[R `TQ@aOTNXF ^ NI_X aOZO@ AZXIQ > OVXLUIS B^[S^NY /@FA > Bfi [ SNUI NI aZPQ^ `NIPUIS [ R`TQQ Y \XOdUUQ_ [R \ OZNINX_O'OOZUIS NIP PUNSZ [ U UI `TQ R'_ NIP_QQ ZP` ^ Y Q_ Q_ [R \ OSZNII OEZ EOQ T``\_!! OXOVZUISZZ aOTNX ^ NI_ZOPaZVI!$ 

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$$\label{eq:started_formula} \begin{split} & \mathsf{FT}[\_Qc\ T[\ aZPQ^{\mathsf{N}}\ MV2 \setminus OZMNXCQ_{\mathsf{T}}\ US \ c\ TQ\ TQ^{\mathsf{N}}\ MV[\ fe[\ faX^{\mathsf{N}}\ U]\ aZPQ^{\mathsf{N}}\ MV2 \ [\ aZP\ aZU_{\mathsf{T}}\ _T[\ aXP\ aZPQ^{\mathsf{N}}\ MV2 \ [\ bQ^{\mathsf{N}}\ MV2 \ U]\ VIP\ Y\ [\ ZU[\ fuZS\ [\ R^{\mathsf{T}}\ QJ \ AVIV_{\mathsf{T}}\ OQQZ\ UZS\ \ faX^{\mathsf{T}}\ _NIP\ \ M^{\mathsf{T}}\ UU_{\mathsf{T}}\ MU2 \ UU_{\mathsf{T}}\ MU2 \ MU2 \$$

 $\begin{aligned} & 3 \text{XX} \text{NTT}[X] & \text{Se XXN}[ ^M[ ^UQ_UZ 3a_ ^ ^ MX0/MQQUbUZ SPaZPUZ S bUM? QPUDMQY a_ NQM00 QPUQP Ne ^TQ \\ & @MUUZ MX3_[ QUMUUZ [ RFQ_UZ S 3a ^T[ ^ UUQ_ /@3F3fi D5B3 > MNI ^M[ ^e 300 QPUMUUZ B^{1} S^{1} XI z z \\ & FTQE` NZPMP_MQ_Q Ne ^ TQ@MUUZ XXBNT[ X] & se 300 QPUMUUZ 3PbU[ ^e 5[ aZQX/@B335fi FTQ \\ & E` NZPMP_MQ_NMQP [ Z ^ TQUZ Q'ZMUUZ NX_ ^ NZPMP ; EA #' #+- E` NZPMP F[ ^? QPUDXX > MNI ^ MI[ ^UQ_Z z z QC LQVXVZP XXNI ^ M[ ^ MQ_MQ NQ0 QPUQP bUM 3@L a_UZ S ^ TQ_NY Q; EA #' #+- M_ ^ TQU^ \\ & NMUUZ T^ \ .!!ccc z' Q NZQPazVA! BMUZ `! > MNZ300 QPUMUUZ fiz \end{aligned}$ 

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 $\begin{array}{l} \label{eq:states} \mathsf{E}[Z[S^{1}\mathsf{M}\mathsf{T}\mathsf{O}^{}](Y]\mathsf{U}SY \mathsf{OPU}\mathsf{O}\mathsf{M}\mathsf{A}X^{1}\mathsf{M}[aZP\,\mathsf{O}\mathsf{O}\mathsf{M}\mathsf{U}\mathsf{U}\mathsf{M}\mathsf{U}Z_Ya_{}^{}] \mathsf{N}_{2}a^{}] \mathsf{N}_{2}a^{}] \mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[aZP\,\mathsf{O}\mathsf{M}\mathsf{M}\mathsf{U}\mathsf{M}\mathsf{U}Z_Ya_{}^{}] \mathsf{N}_{2}a^{}] \mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[aZP\,\mathsf{O}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[Z_Ya_{}^{}] \mathsf{N}_{2}a^{}] \mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[X] \mathsf{M}_{2}a^{}] \mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[X] \mathsf{M}_{2}a^{}] \mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[X] \mathsf{M}_{2}a^{}] \mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[X] \mathsf{M}_{2}a^{}] \mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[X] \mathsf{M}_{2}a^{}] \mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[X] \mathsf{M}_{2}a^{}] \mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}\mathsf{M}[X] \mathsf{M}[X] \mathsf{M$ 

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1.

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 $3 POOKMMUZ [RUZ^OO_RY V _\OORD^[ SalPOXZO_NZP_MOY OZ^_ C M_PODOX(\OP Ne D3@L5A 9)]$ NZIP MI\^[ bCP Ne `TQD3@L5A 9 4[ MP VZ EC\`QY NQ^\$" #\$žFTQ: 9E3!D3@L5A 9 √ VZ` 5[ Y Y U`CQ[ Z  $B^{2}$  MMX6 UMX5 [ U NZIP EO 2021 USY QY NQ^ c Q^ Q a UQP ` [ POOMQ ` TOU^ OXD NZI` UZ Q^ ` UZ  $c UUZS [Z TUR] Y VU ^{(1)} (MEQUAUZS UZ TO ODOC [RTU_TWOY OZ Z$ 

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 $600 \text{ KMZS UZ} 020 \text{ UQ } 02 \text{ UWKUZ } PQ^{(1)} 2 \text{ UWKUZ } RKO \text{ NO C } 02 \text{ TQ} \text{ UWKUZ }$  $U_{2}^{(0)} = [RY QY NQ_{1}^{1} M_{2}^{1} P_{1}^{(0)} Pa_{1}^{(0)} Q_{1}^{(0)} M_{1}^{(0)} [R_{1}^{(0)} Q_{2}^{(0)} Q_{2}^{($ 

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[a`NOX[c UZ 3\\OZPUd 4 \M\* UU052

3` TQ? MOT \$" # + RMODŽ [ŽRMOQ () Y Y U' QQY QQ UZSI TQ QQU UZSI () Z QZ a ŽNMQP  $2 \times 2^{-1}$ NONTRANDON PE [ROUPOZOONZPOXZUDWKOCNO $^{\circ}$ UOZDOQ YYOZPINU Z cO $^{\circ}$ CS $^{\circ}$ MPOPM C

3Z a PMOP XUQ1VIa  $^{O}$  QMOT [ NZ c Q $^{T}$ TQOXZUQXX] aQ UZ c M aZPQ $^{T}$ NX0Zž

 $E^{a}aOa^{OP}OVZUONX[aQ_UZ_cO^{OP}ObOX(OPNZPNS^{OOP}a)[Zž]$ 

`MQYQZ`ž

 $600\text{M/M/UZ} [RUI O'O_ c O'O_ aST R'Y MKY OY NO'_\'U ^ [ 'Obloc UIS TU]$ 

FTU\_`MQY QZ` c M\_[ ^USUZINXX& PODOX[ \ QP UZ 3aSa\_` #--# /5ŽAN\_&FI NZP \_a N\_Q] a QZ` Xe a \ PMQP UZ #--" /5ŽAN\_' fiNZP \$" #' /5ŽAN\_' - fz6a^UZS \$" #%Ž\$" #' ł `TQ: 9E3! D3@L5A9 √ UZ` 5[YY U`QQ[Z  $B^{2}$  MAX6 UNSZ [<u>U</u> MIP EO<sup>2</sup> COZ UIS OM<sup>4</sup> CP [a<sup>\*</sup> TOR] XI c UIS <u>O</u>L UI <sup>4</sup> ColOc UIS TU MOY OZ.

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) QQFOEJY + 7 VIFSNIFX PGU F EFVIFIPION FOUBOE SFWIFX QSPDFTT (PSU JT TUBUFN FOU

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## ) COFOELY, . . VIXII JIDNEUN FS

 $\label{eq:solution} FT \_ UZF[ ^Y MUZ \_ UZ^2CPCP^{[ ^[ buPQSCZCYNXNPbU0D^{[ ^MODUUZC^{+} NZP_T[ a XP Z[ ^ NQ^2XUCP [ Z M_M _ a N_^Ua^2QF[ ^ ^[ ( ^ Q^M_Q Z C UT ^Q_Q QO ^{[ ^TQ M^2U2A} M 00^{-} MZ OQ_[ RONOT OM_QNZP^TQ Z C UT ^Q_Q QO ^{[ ^TQ M^2UA} M 00^{-} MZ OQ_[ RONOT OM_QNZP^TQ Z C UT ^Q_N QD ^{[ ^TQ M^2UA} M 00^{-} MZ OQ_[ RONOT OM_QNZP^TQ Z C UT ^Q_N QD ^{[ ^TQ M^2UA} M 00^{-} MZ OQ_[ RONOT OM_QNZP^TQ Z C UT ^Q_N QD ^{[ ^TQ M^2UA} M 00^{-} MZ OQ_[ RONOT OM_QNZP^TQ Z C UT ^Q_N QD ^{[ ^TQ M^2UA} M 00^{-} MZ OQ_[ RONOT OM_QNZP^TQ Z C UT ^Q_N QD ^{[ ^TQ M^2UA} M 00^{-} MZ OQ_[ RONOT OM_QNZP^{-} MZ OQ_[ RONO$ 

 $\begin{array}{c} \mathsf{TUX}^* \mathsf{TQ5}[\texttt{XCSQQZPQN6}[a^{-}[\texttt{QZ}_a^Q^*\mathsf{TM}\texttt{UZR}^*\texttt{Y}\texttt{MUZ}\texttt{U}\texttt{MODa}^*\mathsf{M}\texttt{Q}\texttt{MZ}^*\texttt{M}^*\mathsf{TQ}^*\texttt{U}\texttt{Q}[\mathsf{R} \\ \land \mathsf{Q}\texttt{M}\mathsf{M}\mathsf{M}\mathsf{U}\mathsf{Z}\mathsf{U}^*\mathsf{U}^*\mathsf{M}\mathsf{Q}\mathsf{Z}\mathsf{U}^*\mathsf{M}\mathsf{Q}\mathsf{U}\mathsf{Z}\mathsf{U}^*\mathsf{M}\mathsf{U}\mathsf{U}\mathsf{Z}\mathsf{U}^*\mathsf{M}\mathsf{Q}\mathsf{U}\mathsf{U}\mathsf{Z}\mathsf{U}^*\mathsf{M}\mathsf{Q}\mathsf{U}\mathsf{U}\mathsf{U}^*\mathsf{U}^*\mathsf{M}\mathsf{Q}\mathsf{U}\mathsf{U}\mathsf{U}^*\mathsf{U}^*\mathsf{M}\mathsf{Q}\mathsf{U}\mathsf{U}\mathsf{U}^*\mathsf{U$ 

) QQFOEJY - +POTJEFSBUIPOT (PS 10EJHFOPVT BOE +VN2V/SBN221 BOE 4JOHVJTUIDBN221, JNFSTF 8PQVN2UIPOT

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   XZSaU\_U00/XKePUbQ\_Q\[\aXX/UZ\_/XZ0APUZSc[YQZR'[YNX];ZPUSQZ[a\_NNX0/XS^[aZPfz]
- &z\$z\$ ;Z 3a\_`^\XXXII`TQ6Q\MYY QZ` [R;Y Y U\$^\VIUZ NZP 5UUGQZ\_TU\ [RRQ^\_8^QQ;Z`Q\ ^Q UZS EQ^bUQQ\_ `T^[ aST F;E @\VIUZ NXRT ^\ \Ub\VIQY OPUDVX\ ^\X0 UUZQ^\_PORZ OP M\_9QZQ^\XXB^\X0 UUZQ^\_NZP ? OPUDVXE\QQUXU\_\_Fi\ ^[ bUPUZS? OPUDMQ ^Q\VIQZ\VIXQ\_QbUQQ\_NZP`TQ.^^QQ\.`UZ Z\_`NRR`[ M^\XISQ NI\[ UZ`Y QZ`\_NZP \ ^[ bUPQ ^Q\_aX\_[ RY OPUDVXCQ\_Z8^QQUZ`Q^\ ^Q Q\_\_MQNXL[ Nb\VXXIVQUZ @Qc LQ\XXZPž

) COFOEJY. + POEJUPOTC WI I JH FSOSFVBNFOODF JO OF PONFIX JI OBSUDVNBSFU OLDUZ

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 $\label{eq:started} FTQR[X][cUZS`NNVQPQ_NUX_`QY_NZP_NNVCbUVNUZ_a_QP`T^[aST[a``TU_`NQY_QZ`žFTQPQRZUUUZ_TNVbQNQQZ`NVVQZ R[Y`TQ@NUUZNX>UV/Me[R? QPUQUZQ_? QPUQUXEaNVQQ: QVPUZS_/? QE: fiPMINNVQQ cTQ'QNVbNXVNVQZ$ 

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7 FTUSIPM	$ \begin{array}{l} AZQ[R^{T}Q^{T}T^{Q}Q^{T}N^{T}Q \\ N[Pe^{Z}; ^{T} \sqcup MT[ ^{T} [ZQ^{T} N^{P}Q Pa^{T}Q^{T}S \land ^{Q}SZ N^{T}Q \\ A^{Q}P^{T} [ ^{T} Q^{T} M^{T} Q N^{T}Q N^{T}Q \\ A^{Q}Q P^{T} [ ^{T} Q^{T} Q N^{T} Q N^{T} Q N^{T} Q \\ M^{T} Q N^{T} Q \\ Q^{T} Q \\ Q^{T} Q \\ Q^{T} Q^{T} Q \\ Q^{T} Q^{T} Q \\ Q^{T} Q^{T} Q^{T} Q \\ Q^{T} Q^{T} Q^{T} Q^{T} Q^{T} \\ Q^{T} Q^{T} Q^{T} \\ Q^{T} Q^{T} Q^{T} \\ Q^{T} Q^{T} Q^{T} Q^{T} Q^{T} \\ Q^{T} Q^{T} Q^{T} Q^{T} \\ Q^{T} Q^{T} Q^{T} \\ Q^{T} Q^{T} \\ Q^{T} Q^{T} \\ Q^{T} Q^{T} Q^{T} \\ Q^{T} Q^{T} Q^{T} \\ Q^{T} Q^{T} \\ Q^{T} \\ Q^{T} Q^{T} \\ Q^{T} \\ Q^{T} Q^{T} \\ Q^{T \\ Q^{T} \\ Q^{T \\ Q^{T} \\ Q^{T \\ Q^{T} \\ Q^{T} \\ Q^{T} \\ Q^{T \\ Q^{T} \\ Q^{T} \\ Q^{T} \\ Q^{T \\ Q^{T} \\ Q^{T \\ Q^{T \\ Q^{T} \\ Q^{T \\ Q^{T} \\ Q^{T} \\ Q^{T \\ Q^{T}$	G7%
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