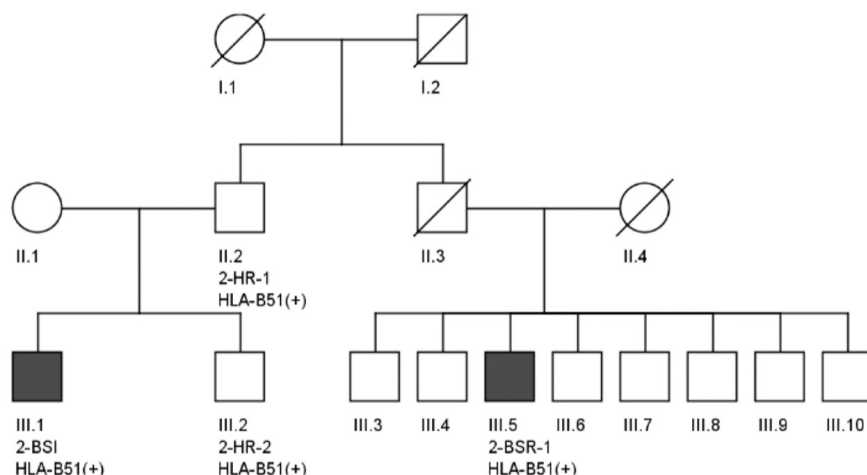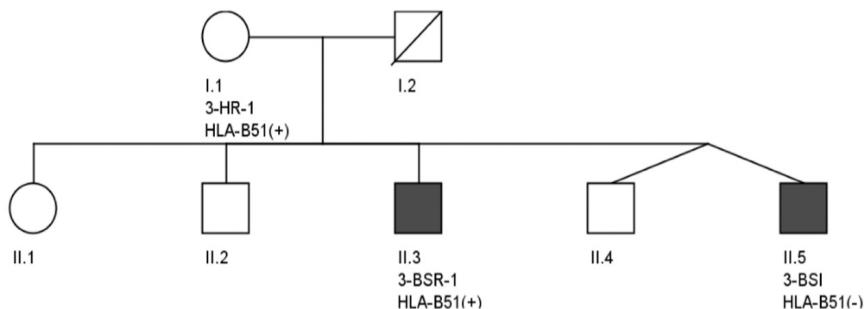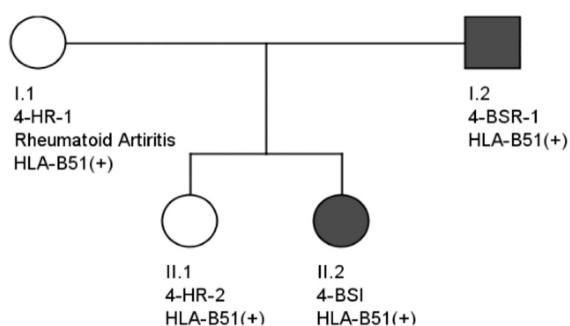**Supplementary Fig. S1.** Pedigree of Family 1. 1-BSI: index patient; HLA-B51(+), 1-BSR-1: first degree relative with BS; HLA-B51(+), 1-HR-1: first degree healthy relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.
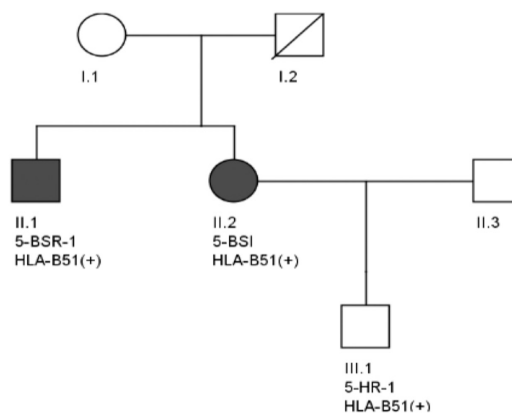

Supplementary Fig. S2. Pedigree of Family 2. 2-BSI: index patient; HLA-B51(+), 2-BSR-1: third degree relative with BS; HLA-B51(+), 2-HR-1: first degree healthy relative; HLA-B51(+), 2-HR-2: first degree healthy relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.

Supplementary Fig. S3. Pedigree of Family 3. 3-BSI: index patient; HLA-B51(-), 3-BSR-1: first degree relative with BS; HLA-B51 (+), 3-HR-1: first degree healthy relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.

Supplementary Fig. S4. Pedigree of Family 4. 4-BSI: index patient; HLA-B51(+), 4-BSR-1: first degree relative with BS; HLA-B51(+), 4-HR-1: first degree relative with RA; HLA-B51(+), 4-HR-2: first degree healthy relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.
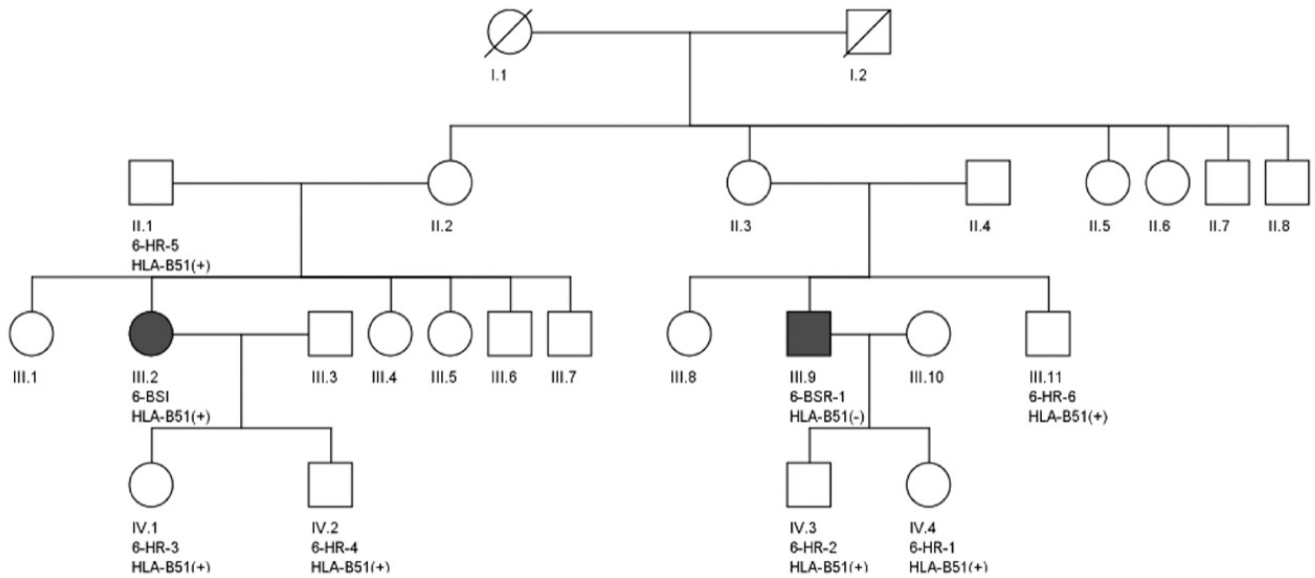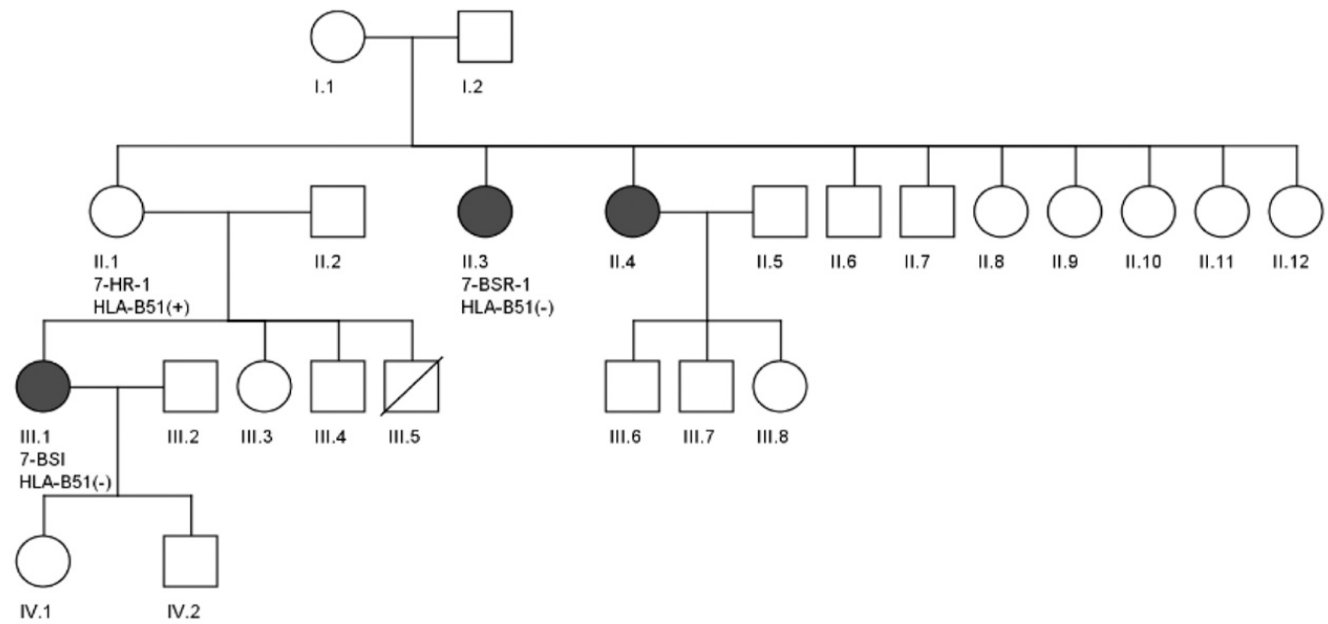

Supplementary Fig. S5. Pedigree of Family 5. 5-BSI: index patient; HLA-B51(+), 5-BSR-1: first degree relative with BS; HLA-B51(+), 5-HR-1: first degree relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.
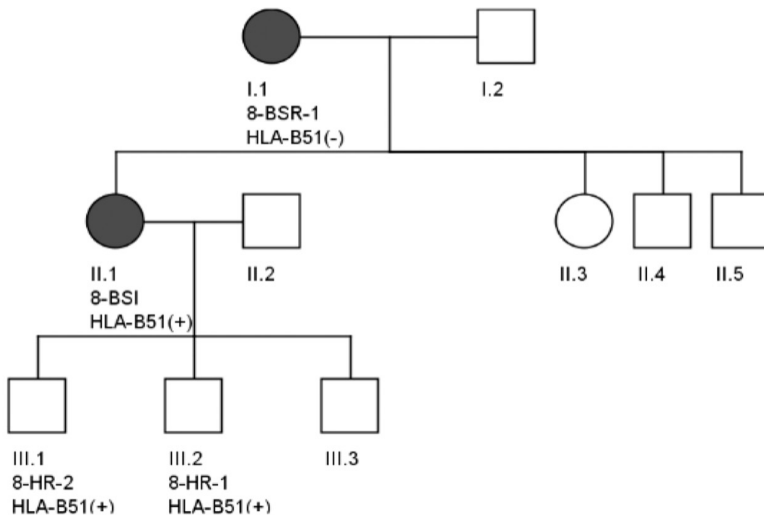
Supplementary Fig. S6. Pedigree of Family 6.
6-BSI: index patient; HLA-B51(+), 6-BSR-1: third degree relative with BS; HLA-B51(-), 6-HR-1; fourth degree healthy relative; HLA-B51(+), 6-HR-2; fourth degree healthy relative; HLA-B51(+), 6-HR-3: first degree healthy relative;HLA-B51(+), 6-HR-4: first degree healthy relative; HLA-B51(+), 6-HR-5: first degree healthy relative; HLA-B51(+), 6-HR-6: third degree healthy relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.
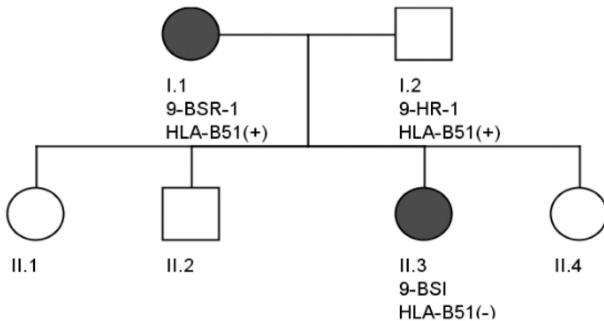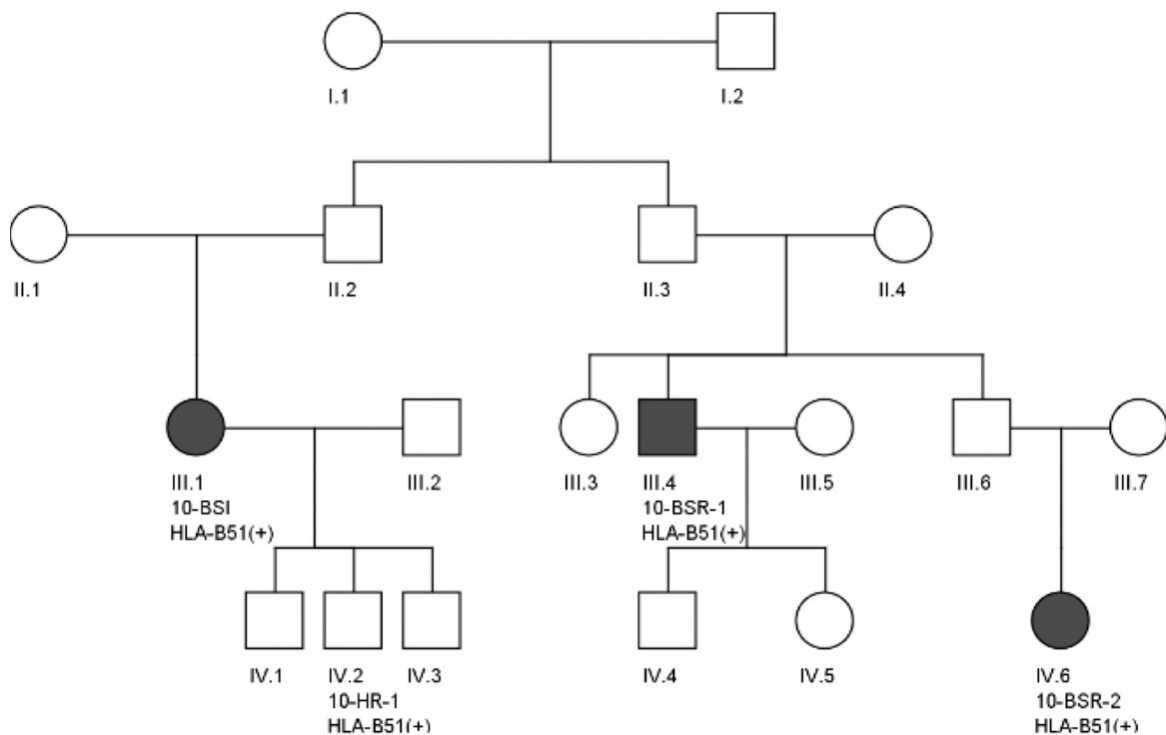

Supplementary Fig. S7. Pedigree of Family 7.
7-BSI: index patient; HLA-B51(-), 7-BSR-1; second degree relative with BS; HLA-B51(-), 7-HR-1: first degree healthy relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.

Supplementary Fig. S8. Pedigree of Family 8. 8-BSI: index patient; HLA-B51(+), 8-BSR-1: first degree relative with BS; HLA-B51(-), 8-HR-1; first degree healthy relative; HLA-B51(+), 8-HR-2: first degree healthy relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.
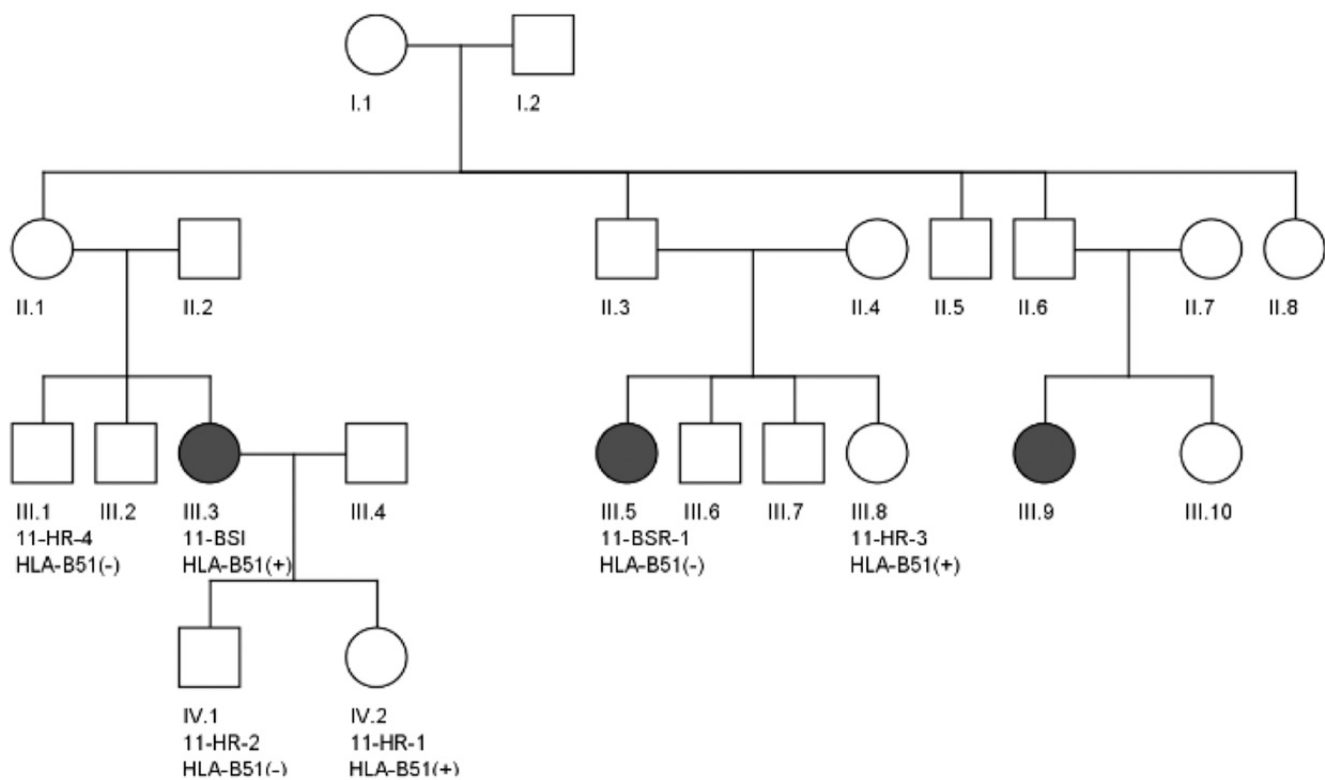

Supplementary Fig. S9. Pedigree of Family 9. 9-BSI: index patient; HLA-B51(-), 9-BSR-1: first degree relative with BS; HLA-B51(+), 9-HR-1: first degree healthy relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.
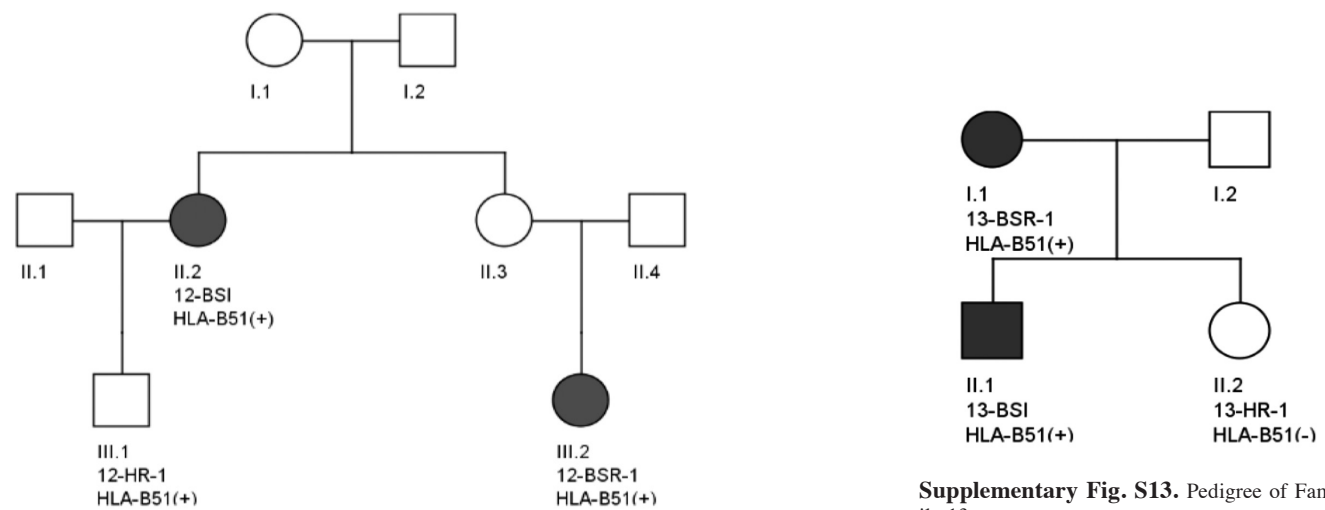

Supplementary Fig. S10. Pedigree of Family 10.
10-BSI: index patient; HLA-B51(+), 10-BSR-1: third degree relative with BS; HLA-B51(+), 10-BSR-2: third degree relative with BS; HLA-B51(+), 10-HR-1: first degree healthy relative; HLA-B51(+) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.

Supplementary Fig. S11. **Pedigree of Family 11.**
11-BSI: index patient; HLA-B51(+), 11-BSR-1: third degree relative with BS; HLA-B51(-), 11-HR-1: first degree healthy relative; HLA-B51(-), 11-HR-2: first degree healthy relative; HLA-B51(-), 11-HR-3: third degree healthy relative; HLA-B51(+), 11-HR-4: first degree healthy relative; HLA-B51(-) Full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.

Supplementary Fig. S12. Pedigree of Family 12.
12-BSI: index patient; HLA-B51(+), 12-BSR-1: second degree relative with; HLA-B51(+), 12-HR-1: first degree healthy relative; HLA-B51(+): full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.
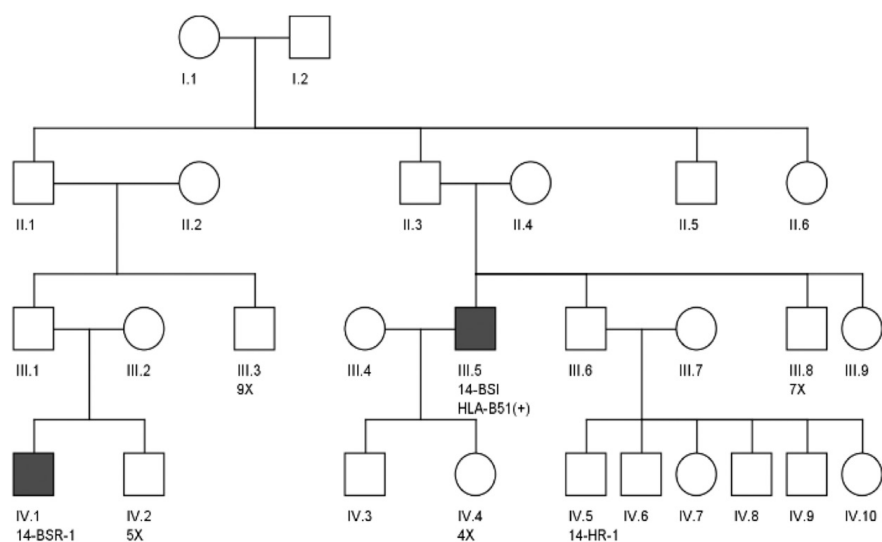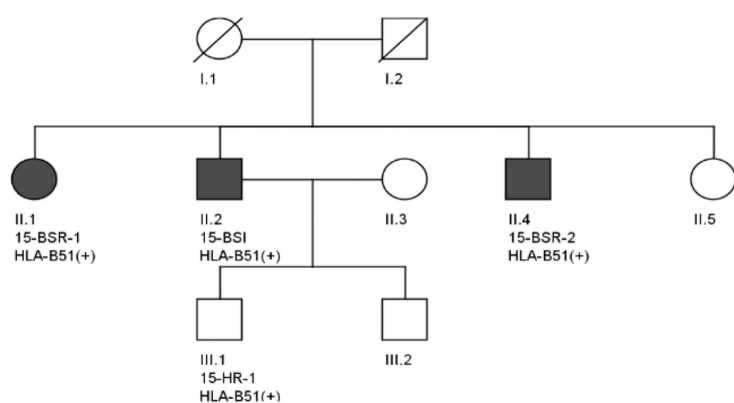

Supplementary Fig. S13. Pedigree of Family 13.
13-BSI: index patient; HLA-B51(+), 13-BSR-1: first degree relative with BS; HLA-B51(+), 13-HR-1: first degree healthy relative; HLA-B51(-): full black filled symbol, clinically proven BS phenotype; open symbol, non-BS.

Supplementary Fig. S14. Pedigree of Family 14.
14-BSI: index patient; HLA-B51(+), 14-BSR-1: fourth degree relative with BS; HLA-B51(+), 14-HR-1: second degree healthy relative; HLA-B51(-) full black filled symbol: clinically proven BS phenotype; open symbol: non-BS.

Supplementary Fig. S15. Pedigree of Family 15.
15-BSI: index patient; HLA-B51(+); 15-BSR-1: first degree relative with BS; HLA-B51(+), 15-BSR-2: first degree relative with BS; HLA-B51(+), 15-HR-1: first degree healthy relative; HLA-B51(+): full black filled symbol, clinically proven BS phenotype; open symbol: non-BS.