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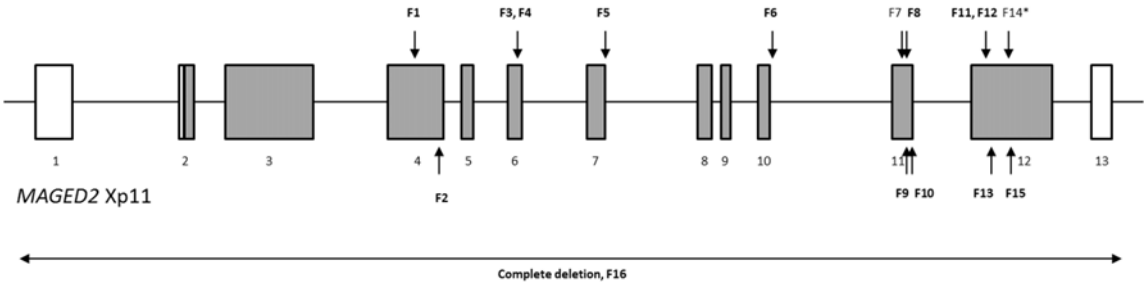
Supplementary Table 1. *In silico* predictions for novel missense mutations

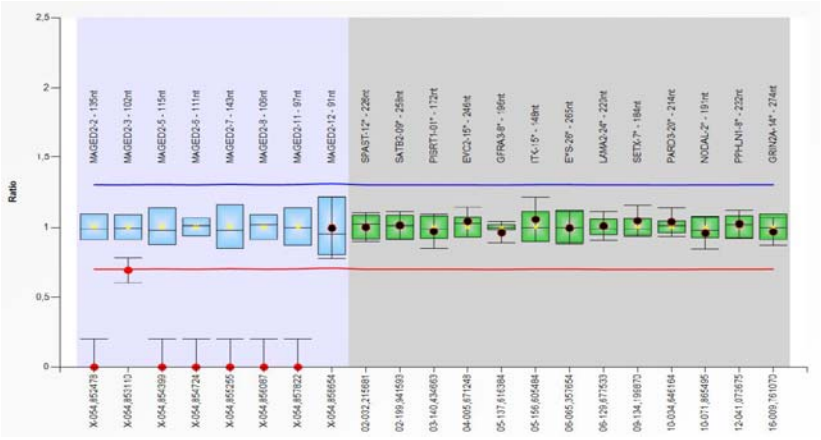
Mutation Nomenclature		**In silico prediction**		
Nucleotide (cDNA)	**Protein**	**SIFT**	**MutationTaster**	**Polyphen-2**
c.1337G>A	p.Arg446His	deleterious (0)	disease causing (0,999)	possibly damaging (0.880)
c.1366G>T	p.Val456Phe	deleterious (0)	disease causing (0,991)	probably damaging (0.928)

Supplementary Figure 1: *MAGED*2 gene structure showing the location of mutations detected. Each number above the exons corresponds to the number of the family in Table 1 and each number below the exons corresponds to the exons' number. The new mutations are in bold. Mutations of families F7 and F14 were previously described. The mutation marked by an asterisk (*) was previously described as c.1462_73del12, p.488_91delEAAA.

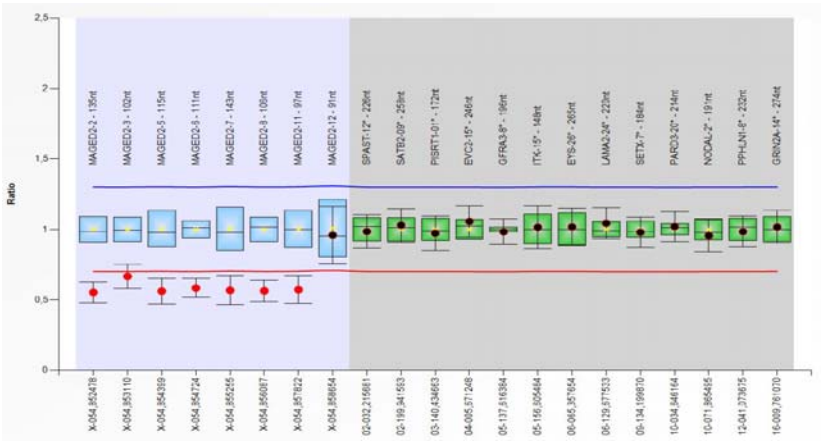


Supplementary Figure 2: MLPA results showing the deletion of *MAGED2* gene: 3A, hemizygous deletion in the proband; 3B, heterozygous deletion in the mother and 3C, only one copy in the father.

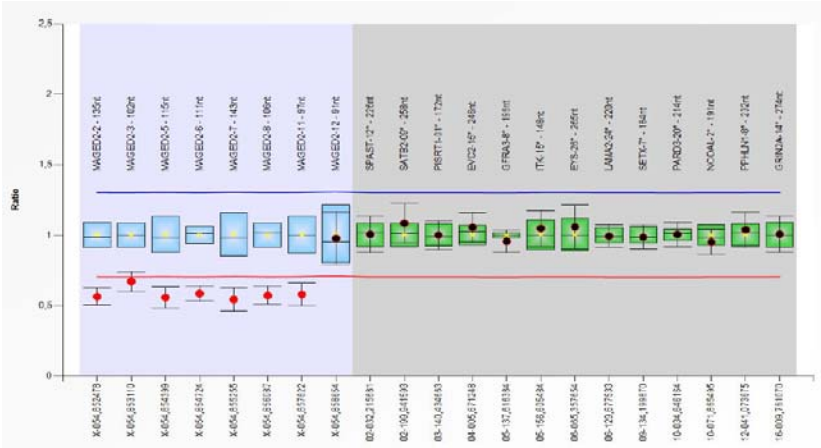
A



B



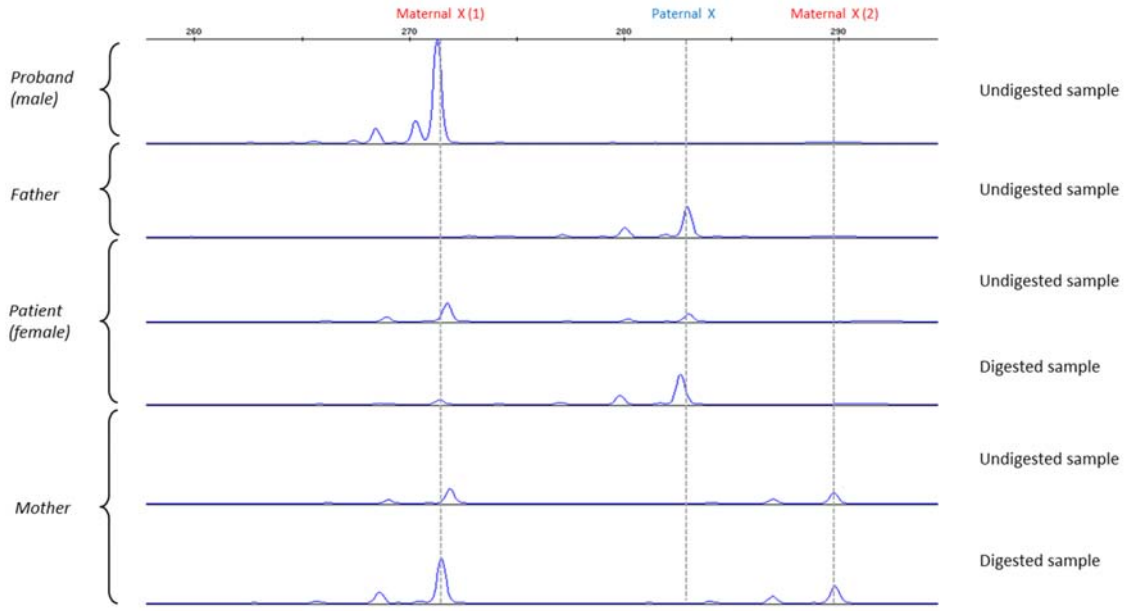
C



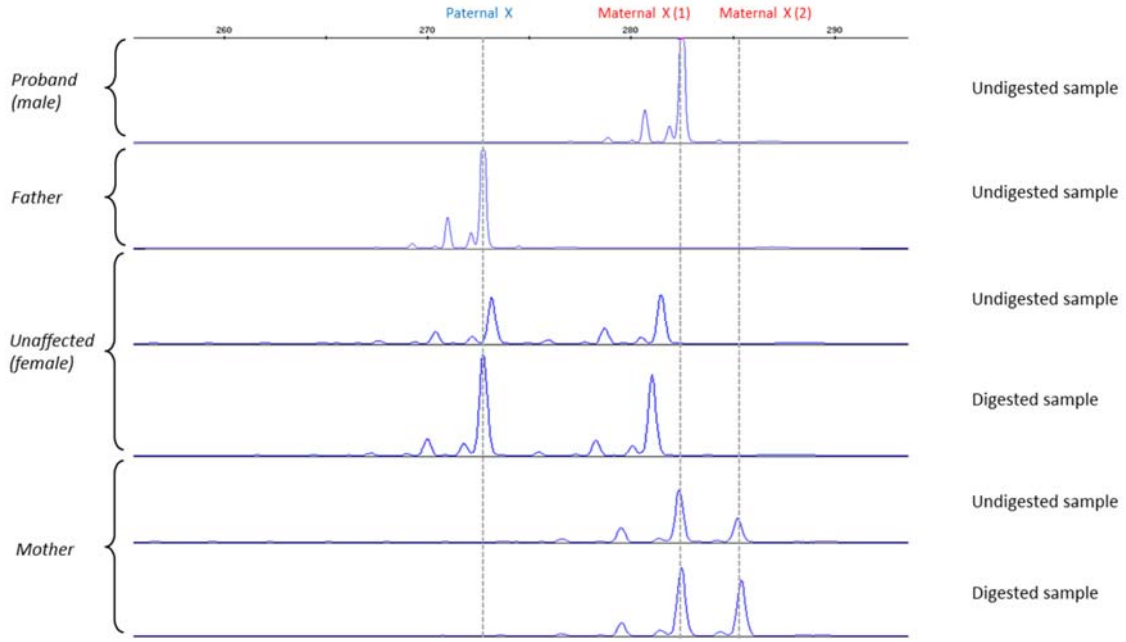
For exon 3, patient and parents have a ratio with ambiguous value (between 0.65 and 0.7). However, the two adjacent exons are deleted and the global profile of the mother is heterozygous, which suggests a hemizygous deletion in the proband. Exon-12 peak has been considered as non-interpretable (i.e., 2 copies for the father).

Supplementary Figure 3: Molecular analysis of X-chromosome inactivation. Examples of two families, one with a bias and the other without. A) Results of X-chromosome inactivation for F2. B) Results of X-chromosome inactivation for F9. The values of relative X inactivation for mutated allele are in red.

A



B



Supplementary Figure 4. Comparison between different types of antenatal Bartter syndrome (*** P<0.0005; **P<0.001; *P<0.005)