Supplementary Table S2

Predictions of bioinformatics tools for variants segregating in families affected with Bardet-Biedl syndrome (BBS)

Family Identity	Genes	Variants	Provean	Sift	Polyphen2,	Mutation taster
LUBS-01 LUBS-02 LUBS-09	BBS6/ MKKS	c.775delA, p.Thr259LeuTer21	Deleterious	Not applicable	Disease causing	Not applicable
LUBS-03	BBS6/ MKKS	c.748G>A, p.gly250Arg	Deleterious	Damaging	Disease causing	Probably Damaging
LUBS-04	BBS9	c.223C>T, p.Arg75Ter	Deleterious	Not applicable	Disease causing	Not applicable
LUBS-05	BBS1	c.1150 C>T, p.Glu384Ter	Stop gain	Deleterious	Not applicable	Disease causing
LUBS-06	BBS2	c.471 +1G>A	Splicing error	Splicing error	Splicing error	Splicing error
LUBS-10	BBS9	c.252delA, p.Lys85SerTer39	Deleterious	Not applicable	Disease causing	Not applicable
CB-03 CB-44	BBS7	c.580delGCA, p.Ala194del	Deleterious	Damaging	Disease causing	Probably Damaging
VI-65	ARL6	c.387_394delAA ATAAAA	Deleterious	Not applicable	Disease causing	Not applicable
RP-04	MKKS	1226G>A, p.Gly409Glu	Deleterious	Damaging	Disease causing	Probably Damaging