SUPPLEMENTARY TABLE 1. Overview of published families with variants in exon 1β.

CDKN2A nucleotide change (exon 1β)†	*CDKN2A* / p14ARF amino acid change	Total no. of families	PDAC reported in families	References
c.45_60dup	p.(Val22Profs*46)	1	Not described	[1]
c.47G>A	p.(Gly16Asp)	1	No	[2]
c.60ins16	insertion	1	No	[3]
c.62G>A	p.(Arg21Lys)	1‡	No	[4]
c.92C>T	p.(Thr31Met)	1	No	[5]
c.81C>G	p.(Ile27Met)	1	Not described	[1]
c.102G>A	p.(Trp34*)	1	Not described	[1]
c.161G>A	p.(Arg54His)	1	No	[6, 7]
c.192A>T	p.(Pro64Pro) (splicing)	1	No	[8, 9]
c.193G>C	p.(Gly65Arg)	8‡, ¶	No (7) Not described (1)	[1, 8, 10-12]
c.193G>A	p.(Gly65Ser)	2	No (1) Not described (1)	[1, 13]
c.193+1G>C	p.? (splicing)	Not described	Not described	[14]
c.193+1G>A	p.? (splicing)	9‡, §	No (8) Not described (1)	[6-9, 12, 15, 16]
c.193+2T>C	p.? (splicing)	1‡	No	[8, 9]
193+3A>G	p.? (splicing)	1‡	No	[8, 9]
c.193+5G>A	p.? (splicing)	2‡	No	[4]
deletion exon 1β	deletion exon 1β	3‡	No	[2, 9, 17]
Total		**35**	**No (29)** **Not described (6)**	

† RefSeq NM_058195.3 isoform p14ARF.
‡ Variant is also reported in Taylor *et al* 2016[18] and Taylor *et al* 2017[14], but the number of families in which the variant was found was not described (and possibly overlaps with other studies).
¶ Includes 4 families that were reported by Potjer *et al* 2019[12] and are included in the current study.
§ Includes 5 families that were reported by Potjer *et al* 2019[12] and are included in the current study.

SUPPLEMENTARY TABLE 2. Overview of published families with variants in exon 2 that solely affect p14ARF.

CDKN2A nucleotide change (exon 2)†	CDKN2A / p16INK4a amino acid change	CDKN2A / p14ARF amino acid change	Total no. of families	PDAC reported in families	References
c.174A>C	p.(Arg58=)	p.(Ser73Arg)	1	Not described	[19-21]
c.192G>A	p.(Leu64=)	p.(Ala79Thr)	1‡	Not described	[14, 19, 22]
c.192G>C	p.(Leu64=)	p.(Ala79Pro)	1	Found in sporadic PDAC patient	[23, 24]
c.198C>T	p.(His66=)	p.(Arg81Trp)	1‡	No	[14, 18, 25]
c.225C>T	p.(Pro75=)	p.(Arg90Cys)	1	Found in sporadic PDAC patient	[24]
c.267C>A	p.(Gly89=)	p.(Leu104Ile)	1	Not described	[22]
c.294C>T	p.(His98=)	p.(Pro113Ser)	2	Not described	[16]
c.306G>A	p.(Ala102=)	p.(Ala117Thr)	1	Not described	[19, 21]
c.318G>A	p.(Val106=)	p.(Ala121Thr)	12‡	Found in sporadic PDAC patient (1) No (4) Not described (7)	[1, 14, 18-24, 26-28]
c.339G>A	p.(Leu113=)	p.(Ala128Thr)	8	Not described (4) No (2)¶ Yes (2)¶	[9, 16, 22, 29-31]
c.339G>C	p.(Leu113=)	p.(Ala128Pro)	Not described	Not described	[14, 18]
Total			**29**	**No (7)** **Not described (17)** **Found in sporadic PDAC (3)** **Yes (2)**	

† RefSeq NM_000077.4 isoform p16INK4a

‡ Variant is also reported in Taylor *et al* 2016[18] and Taylor *et al* 2017[14], but the number of families in which the variant was found was not described (and possibly overlaps with other studies).

¶ In the families reported by Kannengiesser *et al* 2009[30], the variant c.339G>C co-occurred with variant c.340C>T which has an effect on p16INK4a (p.Pro114Ser)

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