# **Request for bed file creation**

Use this form to request the creation of a single BED file (one Pan number). If RPKM and coverage bedfiles are required please submit these on seperate forms.

**Instructions:**

* **Add genes symbols and/or transcripts to a panel in Moka**
* **Add panel description to Moka**
* **Answer following questions:**

1. **Pan number of bed file to be created:**

Pan4301

1. **Panel description/name**

|  |  |  |
| --- | --- | --- |
| **Category** | **SubCategory** | **Panel** |
| VCP2\_Variant**\_**v1 | -30/+30, PTEN\_5UTR-30/+30 +Pan3607,+Pan3609, Pan3611,+Pan4002+ CHEK2\_Chr22:29091857+BRCA\_Chr17:41277375 | VCP2\_Variant\_v1 -30/+30, PTEN\_5UTR-30/+30 +Pan3607,+Pan3609, Pan3611,+Pan4002+ CHEK2\_Chr22:29091857+BRCA\_Chr17:41277375 |

1. **Is this BED file for RPKM?** (delete as appropriate)

No

1. **Is this BED file exclusively based on coordinates provided by a supplier** (e.g SNP identity kits).

No

* 1. If yes, does this need further padding?

N/A

1. **Should specific transcripts be used to create the bed file? (as opposed to all transcripts)** (delete as appropriate)

Yes

|  |  |
| --- | --- |
| APC | NM\_000038 |
| ATM | NM\_000051.3 |
| BMPR1A | NM\_004329 |
| BRCA1 | NM\_007294 |
| BRCA2 | NM\_000059 |
| BRIP1 | NM\_032043 |
| CDH1 | NM\_004360 |
| CHEK2 | NM\_007194.3 |
| EPCAM | NM\_002354 |
| GREM1 | NM\_013372 |
| MLH1 | NM\_000249 |
| MSH2 | NM\_000251 |
| MSH3 | NM\_002439 |
| MSH6 | NM\_000179 |
| MUTYH | NM\_001128425 |
| NBN | NM\_002485 |
| NTHL1 | NM\_002528 |
| PALB2 | NM\_024675 |
| PMS2 | NM\_000535 |
| POLD1 | NM\_001256849 |
| POLE | NM\_006231 |
| PTEN | NM\_000314 |
| RAD51C | NM\_058216 |
| RAD51D | NM\_002878 |
| RNF43 | NM\_017763 |
| SMAD4 | NM\_005359 |
| SMARCA4 | NM\_001128849 |
| STK11 | NM\_000455 |
| TP53 | NM\_000546 |

* 1. **If specific transcripts are to be used have you provided multiple transcripts for the same gene?** (if Yes please state)

No

1. **What padding is required around the exons?**

30bp

1. **Should UTRs be included for ALL genes?** (delete as appropriate)

No

* 1. **Should UTRs be padded?**

N/A

1. **Are UTRs required for SOME genes?** (delete as appropriate)

Yes

* 1. **Please list genes/transcripts.**

PTEN

* 1. **Should UTRs be padded?**

30bp

1. **Any additional BED files to be included in this BED file?** Yes

Please state Pan numbers of additional bed file(s)

Pan3607

Pan3609

Pan3611

Pan4002

1. **Are any further regions required to be added?**

Yes

Please provide genomic coordinates for target regions

Please describe region eg UTR, intronic site.

Does the given regions require padding?

Eg: Chr1:123456-234567 5’ UTR for Gene ABC. Add 10bp padding

CHEK2 Chr22:29091857 single base

Chr17:41277375 BRCA1 c.-107A>T (does not have an rs ID)

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**Requested by: Heidi Kuoppamaa**

**Requested date:** 01/02/2021