# **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:**  
   Pan4302
2. **Panel description/name**

|  |  |  |
| --- | --- | --- |
| **Category** | **SubCategory** | **Panel** |
| VCP1\_Variant\_v1.2 | -30/+30, 5UTR\_LDLR\_PCSK9 +Pan4291,+Pan4290,+,Pan4292,+Pan4272 | VCP1\_Variant\_v1.2 - -10/+10, 5UTR\_LDLR\_PCSK9, +Pan4291,+Pan4290,+,Pan4292,+Pan4272 |

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)

Specific transcripts - see Moka and attached file

|  |  |
| --- | --- |
| ACADM | NM\_000016 |
| ADAMTS13 | NM\_139025 |
| APOB | NM\_000384 |
| APOE | NM\_000041 |
| CFTR | NM\_000492 |
| DMD | NM\_004006 |
| F10 | NM\_000504 |
| F11 | NM\_000128 |
| F13A1 | NM\_000129 |
| F13B | NM\_001994 |
| F2 | NM\_000506 |
| F5 | NM\_000130 |
| F7 | NM\_000131 |
| F8 | NM\_000132 |
| F9 | NM\_000133 |
| FGA | NM\_000508 |
| FGB | NM\_005141 |
| FGFR3 | NM\_000142 |
| FGFR3 | NM\_001163213 |
| FGG | NM\_000509 |
| FGG | NM\_021870 |
| GGCX | NM\_000821 |
| LAMA2 | NM\_000426 |
| LDLR | NM\_000527 |
| LDLRAP1 | NM\_015627 |
| LMAN1 | NM\_005570 |
| MCFD2 | NM\_139279 |
| MTM1 | NM\_000252 |
| MTMR1 | NM\_003828 |
| NOTCH3 | NM\_000435 |
| PCSK9 | NM\_174936 |
| PROC | NM\_000312 |
| PROS1 | NM\_000313 |
| VKORC1 | NM\_024006 |
| VWF | NM\_000552 |

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

Yes

|  |  |
| --- | --- |
| FGFR3 | NM\_000142 |
| FGFR3 | NM\_001163213 |
| FGG | NM\_000509 |
| FGG | NM\_021870 |

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

10bp

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.**

|  |  |
| --- | --- |
| LDLR | NM\_000527 |

|  |  |
| --- | --- |
| PCSK9 | NM\_174936 |

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

5’UTR 10bp

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

Please state Pan Numbers of additional bed file(s)

|  |  |
| --- | --- |
| Haem\_Variant\_v1.1 - Haem SNP Variants | Pan4291 |
| DMD\_Intronic\_Variant\_v1 - Cryptic Splice Variants | Pan4290 |
| FH\_PolySNP\_Variant v1 - PolySNP Variants | Pan4292 |
| CF\_Intronic\_Variant\_v1 | Pan4272 |

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

None

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

**Requested by:** Heidi Kuoppamaa

**Requested date:** 27/01/2021