Introduction to genome annotation - practical information

Enabler for Life Sciences

Karolinska Institutet
KTH
Stockholms universitet
UPPSALA UNIVERSITET
SciLifeLab

Practical info

- Coffee breaks
- Lunch
- Dinner at

  Lingon 19.00

  Svartbäcksg. 30
- Cards

Introduction to genome annotation

What is annotation?

- Identification of regions of interest in sequence data

From a genome…

```
>scaffold_26
AGTCACACACCCTTCAGCTTACACCCTGACTGCAGCCCTTACTCAAAACA
TTCCAGCCAGGAAGATGCTCCGACACAGCTTCTGGATGCCGCTCCTCGAC
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GCCGAGGGCTGGGAGTTATTCCCACACCGGGCCCGTAAGTTTTCTACCCA
AAAACCCATAGAAAAGAGATGAACCACTAAGTTTGATAACTCTTCTACTT
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TGACCGCCGCCTCCTCGCCCGCCACCGCTGAGTACTGCCGTGCCGGAGTC
ACGGAGCCAGTCCCCCGCGGCCCCACCGCCTCCTCGCCCGCCGCCACGGA
GATCGGCTGCGCCACTCCCGAGCTCGGCCGTGCCATCGCCGCCCCCGCCG
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TAAGAAAGCTCAGGCGAATCCCTGCCGTGTCCAAATGCTCTGCTAGAGCA
GGCTGCACCTCGGCAGCATGGCTGTCAGCACAGGGATCAGCAATCAGCGC
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CCAGGGCTCCCCTGCTGCAGGCGCTGCTGAGTGGGACAAGGACATTTTTG
```

…to an annotated gene

GFF file format

```
##gff-version 3
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scaffold_7	maker	mRNA	133848	144662	.	-	.	ID=A649E923246BADE2184E579FA9124ABD;Parent=C55462A8A38E2878A71E2ABDAFB34661;Name=1:cornix-all_reads.72406.1;_AED=1.00;_eAED=1.00;_QI=71|0|0|0|0|0|8|414|347
scaffold_7	maker	exon	138974	139077	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:exon:7;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	exon	135098	135281	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:exon:6;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	exon	139616	139836	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:exon:5;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	exon	144511	144662	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:exon:4;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	exon	136342	136437	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:exon:3;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	exon	133848	134338	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:exon:2;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	exon	141262	141383	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:exon:1;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	exon	144138	144296	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:exon:0;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	five_prime_UTR	144592	144662	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:five_prime_utr;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	CDS	144511	144591	.	-	0	ID=A649E923246BADE2184E579FA9124ABD:cds;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	CDS	144138	144296	.	-	0	ID=A649E923246BADE2184E579FA9124ABD:cds;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	CDS	141262	141383	.	-	0	ID=A649E923246BADE2184E579FA9124ABD:cds;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	CDS	139616	139836	.	-	1	ID=A649E923246BADE2184E579FA9124ABD:cds;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	CDS	138974	139077	.	-	2	ID=A649E923246BADE2184E579FA9124ABD:cds;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	CDS	136342	136437	.	-	0	ID=A649E923246BADE2184E579FA9124ABD:cds;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	CDS	135098	135281	.	-	0	ID=A649E923246BADE2184E579FA9124ABD:cds;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	CDS	134262	134338	.	-	2	ID=A649E923246BADE2184E579FA9124ABD:cds;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	three_prime_UTR	133848	134261	.	-	.	ID=A649E923246BADE2184E579FA9124ABD:three_prime_utr;Parent=A649E923246BADE2184E579FA9124ABD
scaffold_7	maker	gene	83101	117593	.	+	.	ID=D3BD9A5F27797F56A844A2E890FF6B99;Name=maker-scaffold_7-augustus-gene-0.6
scaffold_7	maker	mRNA	83101	117593	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26;Parent=D3BD9A5F27797F56A844A2E890FF6B99;Name=maker-scaffold_7-augustus-gene-0.6-mRNA-1;_AED=1.00;_eAED=1.00;_QI=0|0|0|0|0|0|21|4|706
scaffold_7	maker	exon	95748	95871	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:8;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	99113	99137	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:9;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	90664	90748	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:10;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	110231	110356	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:11;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	113609	113679	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:12;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	94057	94117	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:13;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	84578	84670	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:14;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	115452	115536	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:15;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	111579	111669	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:16;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	102917	103016	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:17;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	96766	96849	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:18;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	86666	86750	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:19;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	99944	100109	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:20;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	109766	109860	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:21;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	93154	93282	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:22;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	114737	114825	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:23;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	83101	83155	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:24;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	108533	108795	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:25;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	117477	117593	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:26;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	106779	106866	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:27;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	exon	105743	105835	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:exon:28;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	83101	83155	.	+	0	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	84578	84670	.	+	2	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	86666	86750	.	+	2	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	90664	90748	.	+	1	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	93154	93282	.	+	0	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	94057	94117	.	+	0	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	95748	95871	.	+	2	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	96766	96849	.	+	1	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	99113	99137	.	+	1	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	99944	100109	.	+	0	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	102917	103016	.	+	2	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	105743	105835	.	+	1	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	106779	106866	.	+	1	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	108533	108795	.	+	0	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	109766	109860	.	+	1	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	110231	110356	.	+	2	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	111579	111669	.	+	2	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	113609	113679	.	+	1	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	114737	114825	.	+	2	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	115452	115536	.	+	0	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	CDS	117477	117589	.	+	2	ID=CFF5DDA190832937C45A0D2E674C9C26:cds;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	three_prime_UTR	117590	117593	.	+	.	ID=CFF5DDA190832937C45A0D2E674C9C26:three_prime_utr;Parent=CFF5DDA190832937C45A0D2E674C9C26
scaffold_7	maker	gene	22451	37514	.	-	.	ID=E1E94A56071E5D7940C93BE70FD79E56;Name=E1E94A56071E5D7940C93BE70FD79E56
scaffold_7	maker	mRNA	22451	37514	.	-	.	ID=2BB3B792BBFCD8B1C7A4070E76500CC5;Parent=E1E94A56071E5D7940C93BE70FD79E56;Name=scaffold_7:22450-37514;_AED=1.00;_eAED=1.00;_QI=3008|0|0|0|0|0|11|762|414
scaffold_7	maker	mRNA	26810	37514	.	-	.	ID=1D629B10725FB97175585BA23623AFA6;Parent=E1E94A56071E5D7940C93BE70FD79E56;Name=maker-scaffold_7-augustus-gene-0.10-mRNA-1;_AED=1.00;_eAED=1.00;_QI=3008|0|0|0|0|0|10|0|400
scaffold_7	maker	exon	26810	27088	.	-	.	ID=1D629B10725FB97175585BA23623AFA6:exon:40;Parent=1D629B10725FB97175585BA23623AFA6
scaffold_7	maker	exon	27950	28090	.	-	.	ID=2BB3B792BBFCD8B1C7A4070E76500CC5:exon:39;Parent=2BB3B792BBFCD8B1C7A4070E76500CC5,1D629B10725FB97175585BA23623AFA6
scaffold_7	maker	exon	33513	33610	.	-	.	ID=2BB3B792BBFCD8B1C7A4070E76500CC5:exon:38;Parent=2BB3B792BBFCD8B1C7A4070E76500CC5,1D629B10725FB97175585BA23623AFA6
maker.gff
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GFF3 file format

Seqid	source	type	start	end	score	strand	phase	attributes
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Chr1	Snap	mRNA	234	3657	.	+	.	ID=gene1.m1; Parent=gene1;
Chr1	Snap	exon	234	1543	.	+	.	ID=gene1.m1.exon1; Parent=gene1.m1;
Chr1	Snap	CDS	577	1543	.	+	0	ID=gene1.m1.CDS1; Parent=gene1.m1;
Chr1	Snap	exon	1822	2674	.	+	.	ID=gene1.m1.exon2; Parent=gene1.m1;
Chr1	Snap	CDS	1822	2674	.	+	2	ID=gene1.m1.CDS2; Parent=gene1.m1;
		start_ codon						Alias, note, ontology_term …
		stop_ codon						

GTF file format

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Sb_20131119_contig_1    Cufflinks       transcript      1522    2095    1000    .       .       gene_id "CUFF.1"; transcript_id "CUFF.1.1"; FPKM "2.6064385494"; frac "1.000000"; conf_lo "0.948975"; conf_hi "3.440036"; cov "4.817376";
Sb_20131119_contig_1    Cufflinks       exon    1522    2095    1000    .       .       gene_id "CUFF.1"; transcript_id "CUFF.1.1"; exon_number "1"; FPKM "2.6064385494"; frac "1.000000"; conf_lo "0.948975"; conf_hi "3.440036"; cov "4.817376";
Sb_20131119_contig_1    Cufflinks       transcript      3626    4118    1000    .       .       gene_id "CUFF.3"; transcript_id "CUFF.3.1"; FPKM "3.1548106029"; frac "1.000000"; conf_lo "0.828669"; conf_hi "3.729011"; cov "8.517668";
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Sb_20131119_contig_1    Cufflinks       transcript      4855    5340    1000    .       .       gene_id "CUFF.5"; transcript_id "CUFF.5.1"; FPKM "7.0235237898"; frac "1.000000"; conf_lo "2.521814"; conf_hi "6.164435"; cov "16.171080";
Sb_20131119_contig_1    Cufflinks       exon    4855    5340    1000    .       .       gene_id "CUFF.5"; transcript_id "CUFF.5.1"; exon_number "1"; FPKM "7.0235237898"; frac "1.000000"; conf_lo "2.521814"; conf_hi "6.164435"; cov "16.171080";
Sb_20131119_contig_1    Cufflinks       transcript      5398    5975    1000    .       .       gene_id "CUFF.4"; transcript_id "CUFF.4.1"; FPKM "3.1706609980"; frac "1.000000"; conf_lo "1.178010"; conf_hi "3.651831"; cov "4.764328";
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Sb_20131119_contig_10   Cufflinks       transcript      954     2795    1000    .       .       gene_id "CUFF.6"; transcript_id "CUFF.6.1"; FPKM "6.8195889357"; frac "1.000000"; conf_lo "5.175059"; conf_hi "7.577765"; cov "16.605534";
Sb_20131119_contig_10   Cufflinks       exon    954     2795    1000    .       .       gene_id "CUFF.6"; transcript_id "CUFF.6.1"; exon_number "1"; FPKM "6.8195889357"; frac "1.000000"; conf_lo "5.175059"; conf_hi "7.577765"; cov "16.605534";
Sb_20131119_contig_1    Cufflinks       transcript      4502    4718    1000    .       .       gene_id "CUFF.2"; transcript_id "CUFF.2.1"; FPKM "37.5296486924"; frac "1.000000"; conf_lo "2.510193"; conf_hi "9.099450"; cov "60.418389";
Sb_20131119_contig_1    Cufflinks       exon    4502    4718    1000    .       .       gene_id "CUFF.2"; transcript_id "CUFF.2.1"; exon_number "1"; FPKM "37.5296486924"; frac "1.000000"; conf_lo "2.510193"; conf_hi "9.099450"; cov "60.418389";
Sb_20131119_contig_1    Cufflinks       transcript      10522   13208   1000    .       .       gene_id "CUFF.23"; transcript_id "CUFF.23.1"; FPKM "55.6377793473"; frac "1.000000"; conf_lo "48.931832"; conf_hi "55.241530"; cov "121.429110";
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Sb_20131119_contig_1    Cufflinks       transcript      13270   14623   1000    .       .       gene_id "CUFF.7"; transcript_id "CUFF.7.1"; FPKM "41.2374406123"; frac "1.000000"; conf_lo "31.982715"; conf_hi "39.274371"; cov "89.788421";
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Sb_20131119_contig_100022       Cufflinks       transcript      3991    4547    1000    .       .       gene_id "CUFF.54"; transcript_id "CUFF.54.1"; FPKM "52.7578565123"; frac "1.000000"; conf_lo "27.382285"; conf_hi "37.895127"; cov "66.397320";
Sb_20131119_contig_100022       Cufflinks       exon    3991    4547    1000    .       .       gene_id "CUFF.54"; transcript_id "CUFF.54.1"; exon_number "1"; FPKM "52.7578565123"; frac "1.000000"; conf_lo "27.382285"; conf_hi "37.895127"; cov "66.397320";
Sb_20131119_contig_100023       Cufflinks       transcript      1097    2009    1000    .       .       gene_id "CUFF.9"; transcript_id "CUFF.9.1"; FPKM "7.4263254644"; frac "1.000000"; conf_lo "4.474632"; conf_hi "7.830606"; cov "16.282075";
Sb_20131119_contig_100023       Cufflinks       exon    1097    2009    1000    .       .       gene_id "CUFF.9"; transcript_id "CUFF.9.1"; exon_number "1"; FPKM "7.4263254644"; frac "1.000000"; conf_lo "4.474632"; conf_hi "7.830606"; cov "16.282075";
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Sb_20131119_contig_100023       Cufflinks       exon    1       123     1000    .       .       gene_id "CUFF.8"; transcript_id "CUFF.8.1"; exon_number "1"; FPKM "6279.6631156093"; frac "1.000000"; conf_lo "42.624813"; conf_hi "69.749694"; cov "8277.399810";
Sb_20131119_contig_100040       Cufflinks       transcript      1       221     1000    .       .       gene_id "CUFF.10"; transcript_id "CUFF.10.1"; FPKM "233.0138642752"; frac "1.000000"; conf_lo "22.182837"; conf_hi "38.203775"; cov "306.188212";
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Sb_20131119_contig_100107       Cufflinks       transcript      2       255     1000    .       .       gene_id "CUFF.11"; transcript_id "CUFF.11.1"; FPKM "29.5909083872"; frac "1.000000"; conf_lo "3.216803"; conf_hi "9.918474"; cov "41.309085";
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Sb_20131119_contig_100107       Cufflinks       transcript      2041    2331    1000    .       .       gene_id "CUFF.12"; transcript_id "CUFF.12.1"; FPKM "18.4360530685"; frac "1.000000"; conf_lo "2.807793"; conf_hi "8.657363"; cov "24.668034";
Sb_20131119_contig_100107       Cufflinks       exon    2041    2331    1000    .       .       gene_id "CUFF.12"; transcript_id "CUFF.12.1"; exon_number "1"; FPKM "18.4360530685"; frac "1.000000"; conf_lo "2.807793"; conf_hi "8.657363"; cov "24.668034";
Sb_20131119_contig_100111       Cufflinks       transcript      21      129     1000    .       .       gene_id "CUFF.14"; transcript_id "CUFF.14.1"; FPKM "3960.0565774823"; frac "1.000000"; conf_lo "5.622026"; conf_hi "18.740088"; cov "4272.822539";
Sb_20131119_contig_100111       Cufflinks       exon    21      129     1000    .       .       gene_id "CUFF.14"; transcript_id "CUFF.14.1"; exon_number "1"; FPKM "3960.0565774823"; frac "1.000000"; conf_lo "5.622026"; conf_hi "18.740088"; cov "4272.822539";
Sb_20131119_contig_100121       Cufflinks       transcript      1756    2236    1000    .       .       gene_id "CUFF.17"; transcript_id "CUFF.17.1"; FPKM "52.3538569152"; frac "1.000000"; conf_lo "23.640043"; conf_hi "34.398386"; cov "75.482038";
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Sb_20131119_contig_100192       Cufflinks       transcript      1840    2212    1000    .       .       gene_id "CUFF.20"; transcript_id "CUFF.20.1"; FPKM "24.9098132799"; frac "1.000000"; conf_lo "7.484312"; conf_hi "14.786080"; cov "49.916331";
Sb_20131119_contig_100192       Cufflinks       exon    1840    2212    1000    .       .       gene_id "CUFF.20"; transcript_id "CUFF.20.1"; exon_number "1"; FPKM "24.9098132799"; frac "1.000000"; conf_lo "7.484312"; conf_hi "14.786080"; cov "49.916331";
Sb_20131119_contig_100107       Cufflinks       transcript      430     902     1000    .       .       gene_id "CUFF.13"; transcript_id "CUFF.13.1"; FPKM "25.0500552879"; frac "1.000000"; conf_lo "10.508448"; conf_hi "18.137870"; cov "45.523657";
Sb_20131119_contig_100107       Cufflinks       exon    430     902     1000    .       .       gene_id "CUFF.13"; transcript_id "CUFF.13.1"; exon_number "1"; FPKM "25.0500552879"; frac "1.000000"; conf_lo "10.508448"; conf_hi "18.137870"; cov "45.523657";
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Sb_20131119_contig_100192       Cufflinks       exon    1       616     1000    .       .       gene_id "CUFF.19"; transcript_id "CUFF.19.1"; exon_number "1"; FPKM "42.2951435419"; frac "1.000000"; conf_lo "23.433223"; conf_hi "33.823425"; cov "88.806988";
Sb_20131119_contig_10011        Cufflinks       transcript      219     353     1000    .       .       gene_id "CUFF.15"; transcript_id "CUFF.15.1"; FPKM "4118.4602721559"; frac "1.000000"; conf_lo "53.966826"; conf_hi "84.228598"; cov "5420.068745";
Sb_20131119_contig_10011        Cufflinks       exon    219     353     1000    .       .       gene_id "CUFF.15"; transcript_id "CUFF.15.1"; exon_number "1"; FPKM "4118.4602721559"; frac "1.000000"; conf_lo "53.966826"; conf_hi "84.228598"; cov "5420.068745";
Sb_20131119_contig_100040       Cufflinks       transcript      945     2276    1000    -       .       gene_id "CUFF.38"; transcript_id "CUFF.38.1"; FPKM "69.4702369875"; frac "1.000000"; conf_lo "60.275496"; conf_hi "70.712087"; cov "143.103653";
Sb_20131119_contig_100040       Cufflinks       exon    945     1966    1000    -       .       gene_id "CUFF.38"; transcript_id "CUFF.38.1"; exon_number "1"; FPKM "69.4702369875"; frac "1.000000"; conf_lo "60.275496"; conf_hi "70.712087"; cov "143.103653";
Sb_20131119_contig_100040       Cufflinks       exon    2079    2276    1000    -       .       gene_id "CUFF.38"; transcript_id "CUFF.38.1"; exon_number "2"; FPKM "69.4702369875"; frac "1.000000"; conf_lo "60.275496"; conf_hi "70.712087"; cov "143.103653";
Sb_20131119_contig_100121       Cufflinks       transcript      1       150     1000    .       .       gene_id "CUFF.16"; transcript_id "CUFF.16.1"; FPKM "1080.5400540118"; frac "1.000000"; conf_lo "23.150255"; conf_hi "44.484805"; cov "1391.311243";
Sb_20131119_contig_100121       Cufflinks       exon    1       150     1000    .       .       gene_id "CUFF.16"; transcript_id "CUFF.16.1"; exon_number "1"; FPKM "1080.5400540118"; frac "1.000000"; conf_lo "23.150255"; conf_hi "44.484805"; cov "1391.311243";
Sb_20131119_contig_10022        Cufflinks       transcript      1510    2616    1000    .       .       gene_id "CUFF.22"; transcript_id "CUFF.22.1"; FPKM "10.9257515912"; frac "1.000000"; conf_lo "7.442428"; conf_hi "11.194395"; cov "24.833916";
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Sb_20131119_contig_100112       Cufflinks       transcript      933     3862    1000    .       .       gene_id "CUFF.21"; transcript_id "CUFF.21.1"; FPKM "13.6659124939"; frac "1.000000"; conf_lo "11.688997"; conf_hi "14.524101"; cov "32.588772";
Sb_20131119_contig_100112       Cufflinks       exon    933     3862    1000    .       .       gene_id "CUFF.21"; transcript_id "CUFF.21.1"; exon_number "1"; FPKM "13.6659124939"; frac "1.000000"; conf_lo "11.688997"; conf_hi "14.524101"; cov "32.588772";
Sb_20131119_contig_10022        Cufflinks       transcript      2857    6365    1000    .       .       gene_id "CUFF.24"; transcript_id "CUFF.24.1"; FPKM "13.7863265713"; frac "1.000000"; conf_lo "12.049946"; conf_hi "14.805328"; cov "32.698360";
Sb_20131119_contig_10022        Cufflinks       exon    2857    6365    1000    .       .       gene_id "CUFF.24"; transcript_id "CUFF.24.1"; exon_number "1"; FPKM "13.7863265713"; frac "1.000000"; conf_lo "12.049946"; conf_hi "14.805328"; cov "32.698360";
Sb_20131119_contig_100080       Cufflinks       transcript      39      1611    1000    -       .       gene_id "CUFF.18"; transcript_id "CUFF.18.1"; FPKM "44.1667134200"; frac "1.000000"; conf_lo "34.717689"; conf_hi "42.267104"; cov "77.065569";
Sb_20131119_contig_100080       Cufflinks       exon    39      135     1000    -       .       gene_id "CUFF.18"; transcript_id "CUFF.18.1"; exon_number "1"; FPKM "44.1667134200"; frac "1.000000"; conf_lo "34.717689"; conf_hi "42.267104"; cov "77.065569";
Sb_20131119_contig_100080       Cufflinks       exon    207     1041    1000    -       .       gene_id "CUFF.18"; transcript_id "CUFF.18.1"; exon_number "2"; FPKM "44.1667134200"; frac "1.000000"; conf_lo "34.717689"; conf_hi "42.267104"; cov "77.065569";
Sb_20131119_contig_100080       Cufflinks       exon    1128    1611    1000    -       .       gene_id "CUFF.18"; transcript_id "CUFF.18.1"; exon_number "3"; FPKM "44.1667134200"; frac "1.000000"; conf_lo "34.717689"; conf_hi "42.267104"; cov "77.065569";
Sb_20131119_contig_10026        Cufflinks       transcript      1       1002    1000    .       .       gene_id "CUFF.49"; transcript_id "CUFF.49.1"; FPKM "49.5933867311"; frac "1.000000"; conf_lo "35.335602"; conf_hi "43.965643"; cov "84.354408";
Sb_20131119_contig_10026        Cufflinks       exon    1       1002    1000    .       .       gene_id "CUFF.49"; transcript_id "CUFF.49.1"; exon_number "1"; FPKM "49.5933867311"; frac "1.000000"; conf_lo "35.335602"; conf_hi "43.965643"; cov "84.354408";
Sb_20131119_contig_10031        Cufflinks       transcript      26      377     1000    .       .       gene_id "CUFF.25"; transcript_id "CUFF.25.1"; FPKM "43.3120795956"; frac "1.000000"; conf_lo "12.379816"; conf_hi "22.244981"; cov "78.396470";
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Sb_20131119_contig_10032        Cufflinks       transcript      5174    5386    1000    .       .       gene_id "CUFF.29"; transcript_id "CUFF.29.1"; FPKM "386.7080614787"; frac "1.000000"; conf_lo "33.884660"; conf_hi "52.744990"; cov "584.846045";
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Sb_20131119_contig_10032        Cufflinks       transcript      160     3305    1000    .       .       gene_id "CUFF.35"; transcript_id "CUFF.35.1"; FPKM "5.9167626598"; frac "1.000000"; conf_lo "4.912969"; conf_hi "6.795913"; cov "12.795227";
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Sb_20131119_contig_100403       Cufflinks       transcript      822     1053    1000    .       .       gene_id "CUFF.30"; transcript_id "CUFF.30.1"; FPKM "90.3398746415"; frac "1.000000"; conf_lo "9.098097"; conf_hi "19.663630"; cov "118.213868";
Sb_20131119_contig_100403       Cufflinks       exon    822     1053    1000    .       .       gene_id "CUFF.30"; transcript_id "CUFF.30.1"; exon_number "1"; FPKM "90.3398746415"; frac "1.000000"; conf_lo "9.098097"; conf_hi "19.663630"; cov "118.213868";
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Sb_20131119_contig_10031        Cufflinks       transcript      1415    3072    1000    .       .       gene_id "CUFF.36"; transcript_id "CUFF.36.1"; FPKM "55.9644866986"; frac "1.000000"; conf_lo "45.789638"; conf_hi "53.715558"; cov "119.323264";
Sb_20131119_contig_10031        Cufflinks       exon    1415    3072    1000    .       .       gene_id "CUFF.36"; transcript_id "CUFF.36.1"; exon_number "1"; FPKM "55.9644866986"; frac "1.000000"; conf_lo "45.789638"; conf_hi "53.715558"; cov "119.323264";
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Sb_20131119_contig_100328       Cufflinks       transcript      1505    3535    1000    .       .       gene_id "CUFF.33"; transcript_id "CUFF.33.1"; FPKM "4.5481695975"; frac "1.000000"; conf_lo "3.453060"; conf_hi "5.296928"; cov "10.333563";
transcripts.gtf
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Why is annotation important?

There are two major parts of annotation

- 1) Structural: Find out where the regions of interest (usually genes) are in the genome and what they look like. How many exons/introns? UTRs? Isoforms?

- 2) Functional: Find out what the regions do. What do they code for?

Open reading frames

Anonymous

Frame	from	to	Length
+3	1383	1736	354
+3	2511	2756	246
-2	2465	2686	222
-1	2880	3092	213
-2	2996	3187	192
-3	574	753	180
+2	2753	2929	177
-2	2	172	171
-1	2034	2171	138
+3	3237	3349	114
+3	2874	2984	111
-2	1283	1393	111
+1	1345	1446	102
-2	599	700	102

Difficult in practice

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Combine data - use Maker!

- External data - proteins, rna-seq (incl. ESTs)
- Ab-initio gene finders
- (Lift-overs from closely related genomes)

MAKER
Annotate this!

Combined annotation

Data used - Proteins

Data used - Proteins

- Conserved in sequence => conserved annotation with little noise
- Proteins from model organisms often used => bias?
- Proteins can be incomplete => problems as many annotation procedures are heavily dependent on protein alignments

```
>ENSTGUP00000017616 pep:novel chromosome:taeGut3.2.4:8_random:2849599:2959678:-1 gene:ENSTGUG00000017338 transcript:ENSTGUT000000180
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>ENSTGUP00000017615 pep:novel chromosome:taeGut3.2.4:23_random:205321:209117:1 gene:ENSTGUG00000017337 transcript:ENSTGUT00000018017
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RLKPRIPAEGDCPPEDSGEERSSPCDLTFEDIMPAVKTLIRAVRILKFLVAKRKFKETLR
PYDVKDVIEQYSAGHLDMLGRIKSLQTRVEQIVGRDRALPADKKVREKGEKPALEAELVD
ELSMMGRVVKVERQVQSIEHKLDLLLGLYSRCLRKGSANSLVLAAVRVPPGEPDVTSDYQ
SPVEHEDISTSAQSLSISRLASTNMD
```

Data used - Proteins

- Maker will align proteins for you: Blast -> Exonerate
- Blast is not structure aware, Exonerate is (splice sites, start/stop codons)
- Preferred file-format: fasta

RNA-seq

DNA
Exon
Intron
Exon
Intron
Exon
Intron
Exon
UTR
UTR
ATG
Start codon
GT
AG
GT
AG
GT
AG
TAG, TAA, TGA
Stop codon
Transcription
Pre-mRNA
UTR
UTR
AA
A
A
A
A
A
ATG
Start codon
TAG, TAA, TGA
Stop codon
Splicing
mRNA
UTR
UTR
AAAAAAAAA
ATG
Start codon
TAG, TAA, TGA
Stop codon
Translation

Data used - RNA-seq

- Should always be included in an annotation project
- From the same organism as the genomic data => unbiased
- Can be very noisy (tissue/species dependent), can include pre-mRNA
- PASA, or some other filtering method, often needed

Spliced reads

RNA-seq - Spliced reads

Pre-mRNA

Pre-mRNA

DNA
Exon
Intron
Exon
Intron
Exon
Intron
Exon
UTR
UTR
ATG
Start codon
GT
GT
GT
TAG, TAA, TGA
Stop codon
Transcription
Pre-mRNA
UTR
UTR
ATG
Start codon
TAG, TAA, TGA
Stop codon
Splicing
mRNA
UTR
UTR
ATG
Start codon
TAG, TAA, TGA
Stop codon
Translation

Pre-mRNA

7,730 kb
7,740 kb
7,750 kb
7,760 kb
7,770 kb
CUFF.28035.1
CUFF.27994.1
CUFF.28031.4
CUFF.28011.1
CUFF.28042.1
CUFF.28031.2
CUFF.28013.1
CUFF.28042.2
CUFF.28031.3
CUFF.28031.1
CUFF.28031.5

Includes everything that is transcribed

Includes everything that is transcribed

Stranded rna-seq

Three-prime bias in polyA-selected rna-seq

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How to use RNA-seq

- Maker will align transcripts (ESTs), but these need to be assembled first.
- Cufflinks: mapped reads -> transcripts

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Mapped Trinity-assembled transcripts

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How to use RNA-seq

- Maker will align transcripts (ESTs), but these need to be assembled first.
- Cufflinks: mapped reads -> transcripts
- Trinity: assembles transcripts without a genome
- PASA can be used to improve transcript quality

Ab initio gene finders are used in Maker

- Commonly used programs: Augustus, Snap, Genemark-ES, FGENESH, Genscan, Glimmer-HMM,…
- Uses HMM-models to figure out how introns, exons, UTRs etc. are structured
- These HMM-models need to be trained!

Liftovers are very useful for orthology determination

- Kraken
- Align the two genomes (Satsuma) and then transfer annotations between aligned regions

General recommendations

- Always combine different types of evidence!
- One single method is not enough!
- Use Maker!

MAKER
Annotate this!

Or get help - BILS annotation resource

- Three people 100% working with annotation
- Deliver high quality annotations
- Enable visualization and manual curation through a web interface
- Also available for consultation

Biosupport.se

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questions

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