

CSE 182 Final Presentation

GROUP 7: Kayla Cooper, Riani Shah, Sameera Agarwala, Dhruv Patravali, Ryan Izad

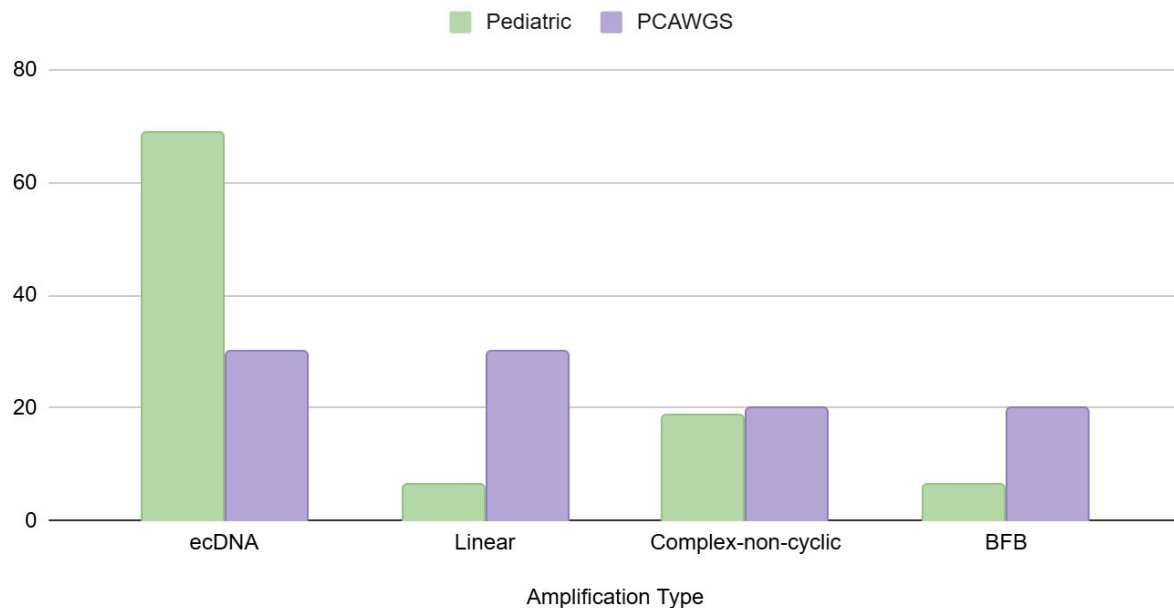
Part I: CCND2 Amplification Types

- Filtered samples containing CCND2
- Pediatric dataset: 16 samples
- PCAWG (Pan-Cancer Analysis of Whole Genomes): 10 samples
- Counted and compared amplification types



Part I: Amplification Types

% of Amplification Types for Samples Containing CCND2 (Pediatric vs PCAWG)

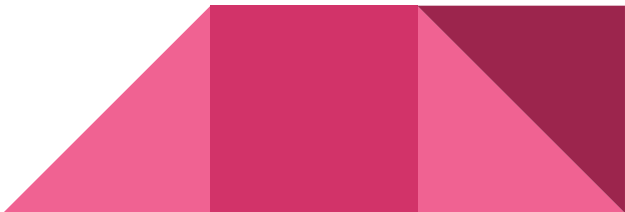


Part I: Age-Based Pathology

In children: Associated with neurodevelopmental disorders like:

- Microcephaly
- MPPH Syndrome (brain overgrowth, cortical abnormalities, polydactyly)

In adults:

- Linked to reproductive tumors (ovarian/testicular)
 - Correlated with poor prognosis in adult gliomas
- 

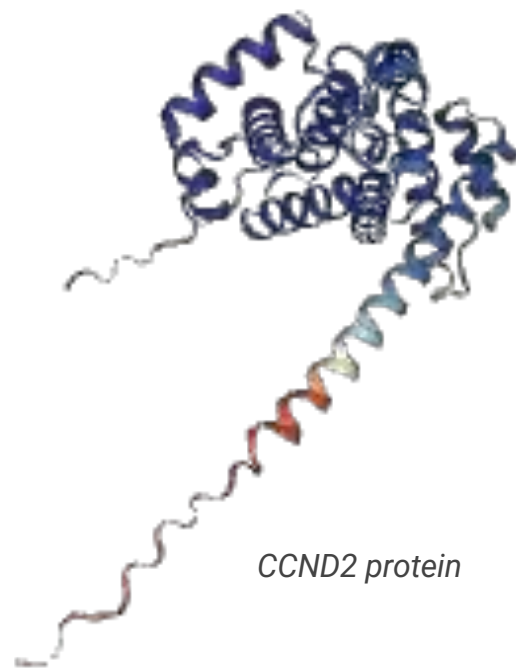
Part I: CCND2-CDK4/CDK6 Interactions

- CCND2 forms complexes with **CDK4/CDK6** to regulate Rb protein
- Drives **cell cycle progression** (G1/S transition)
- Genomic location:
 - CCND2**: Chr 12p13.32
 - CDK4**: Chr 12q14.1
 - CDK6**: Chr 7q21.2
- CDK4 and CCND2 more likely to **co-amplify**



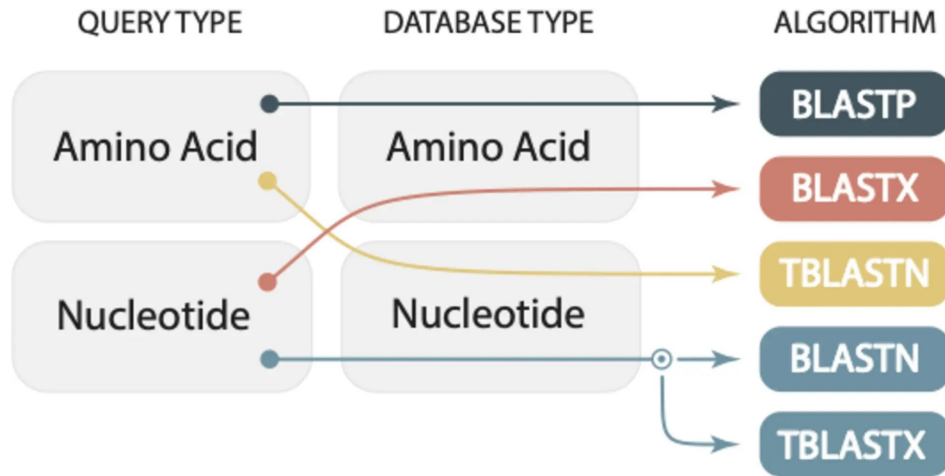
Part II

1. Extract Protein Sequences
 - Retrieved Cyclin D2 Protein (NCBI RefSeq)
 - Encoded by CCND2 oncogene
2. Extract Genomic Region from ecDNA
 - BED file from Amplicon Repository
 - Chr12 intervals for ecDNA
 - bedtools for fasta
3. Blast protein against ecDNA



Part II: tblastn

- Blast tool: protein seq to db query
- Searches all 6 reading frames



Part II

Query ID	Subject ID	% Identity	Alignment Length	Mismatches	Gap Opens	Query Start	Query End	Subject Start	Subject End	E-value	Bit Score
NP_001750.1	chr12:874783-4459229	100.000	61	0	0	77	137	3510421	3510603	1.55e-33	127
NP_001750.1	chr12:874783-4459229	96.429	56	2	0	136	191	3513137	3513304	2.45e-29	114
NP_001750.1	chr12:874783-4459229	88.333	60	7	0	181	240	3523194	3523373	7.63e-26	104
NP_001750.1	chr12:874783-4459229	100.000	49	0	0	241	289	3534243	3534389	2.71e-24	100
NP_001750.1	chr12:874783-4459229	100.000	64	0	0	1	64	3508424	3508615	1.99e-21	91.7

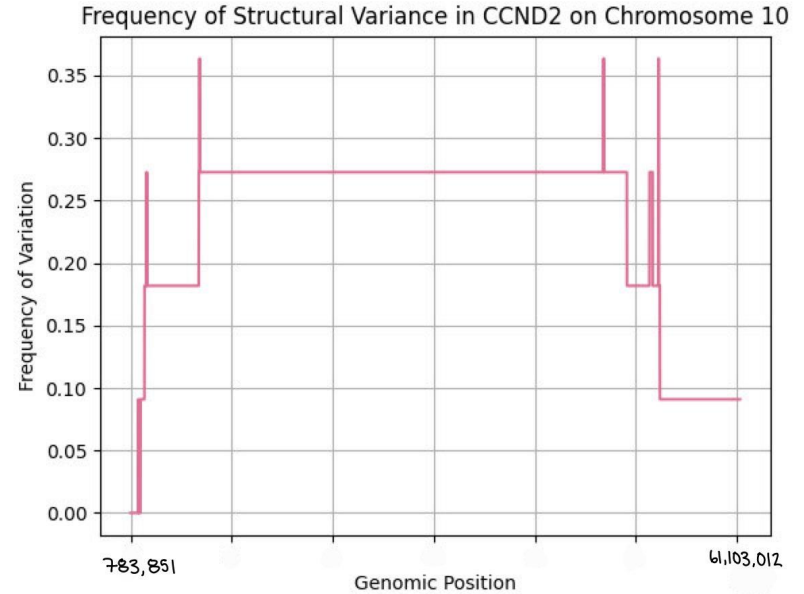
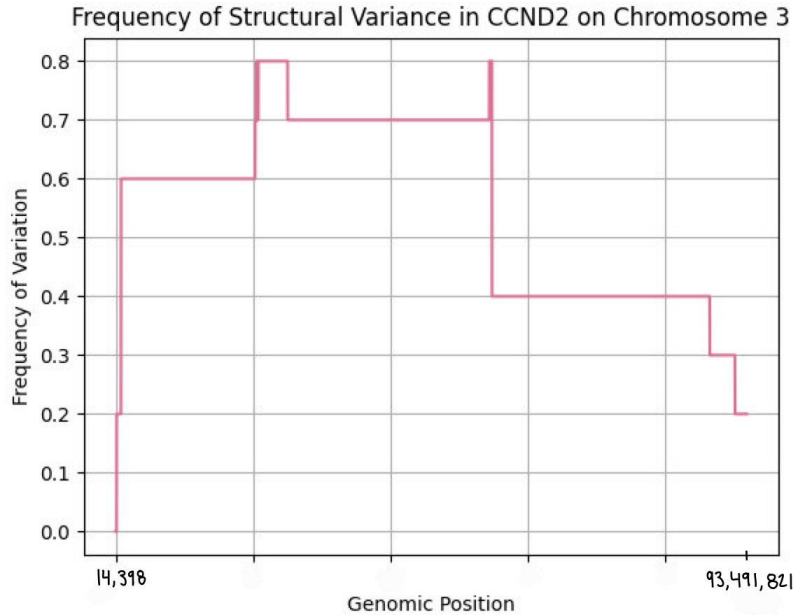
- Full protein detected (amino acids 1–289) → no truncation
- Alignments span multiple ecDNA regions → gene is fragmented
- High identity across segments (100%, 96%, 88%) → gene likely functional.
- Decreasing identity (88%) in one region suggests possible mutation or rearrangement
- Supports ecDNA's role in preserving oncogenes despite structural scrambling

Part III

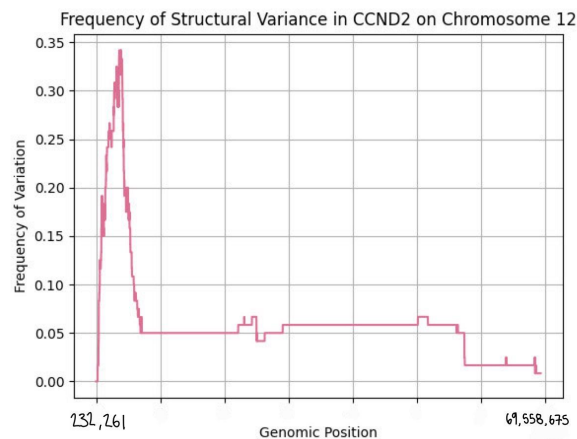
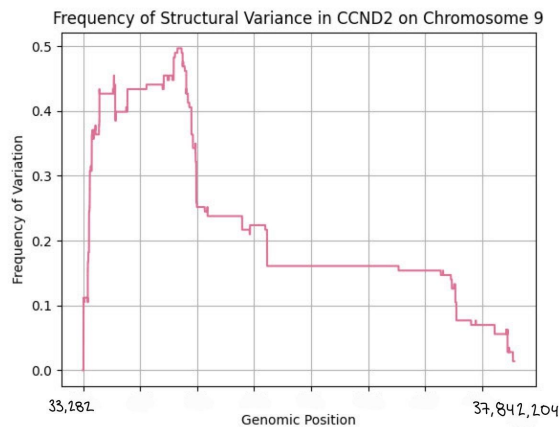
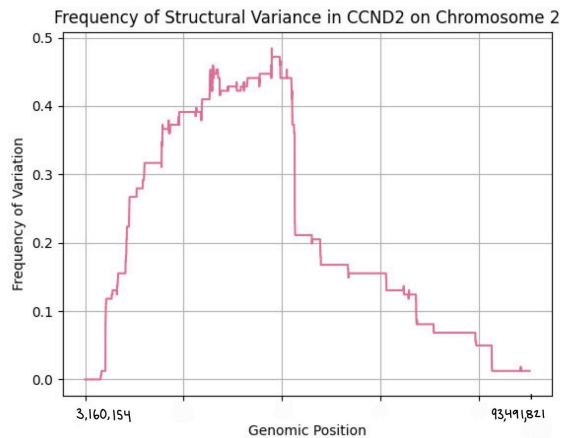
1	discordant	chr12:63953228+>chr12:3998332-	11.956269348671515	62	None	None
2	discordant	chr12:68421563+>chr12:63952767-	11.96028250867962	86	None	None
3	discordant	chr12:68425775->chr12:68404838-	12.367406327413487	73	None	None
4	discordant	chr12:68600914->chr12:68547789-	4.755363685379145	31	None	None
5	discordant	chr12:68714179+>chr8:38034549-	4.369114798841769	27	None	None
6	discordant	chr12:69558675+>chr12:5101439+	12.065089355129132	54	None	None
7	discordant	chr12:359986->chr12:359986-	2.834484274053305	14	None	None
8	discordant	chr12:405365+>chr12:404493-	1.319441472512009	6	None	None
9	discordant	chr12:920419->chr12:919663+	3.6579670950888	18	None	None
10	discordant	chr12:966072+>chr12:964881+	0.0577969526558102	5	None	None
11	discordant	chr12:1329444->chr12:1328825+	3.8081476729703763	39	None	None
12	discordant	chr12:1588089+>chr12:232261+	0.12628314574130461	5	None	None
13	discordant	chr12:2106410->chr12:2105669+	1.8881273593197452	8	None	None
14	discordant	chr12:2193814+>chr12:729595+	0.2862583449629201	7	None	None
15	discordant	chr12:2289865->chr12:2289330+	2.688897898296888	2	None	None
16	discordant	chr12:2382366->chr12:1689725-	0.4824411605274848	6	None	None
17	discordant	chr12:2385634+>chr12:1925377+	1.0672929294877114	3	None	None
18	discordant	chr12:2750761+>chr12:2749184+	0.8834864529611675	9	None	None
19	discordant	chr12:3017115->chr12:945731-	1.1281191489310416	7	None	None
20	discordant	chr12:4244000->chr12:379682-	78.36576265797176	485	None	None
21	discordant	chr12:4656962->chr12:2299679+	9.487490261572303	79	None	None
22	discordant	chr12:4936412+>chr12:3722192-	0.24036901664108645	6	None	None
23	discordant	chr12:5079212->chr12:2740811+	1.1503571290368888	5	None	None
24	discordant	chr12:5213172+>chr12:4858286-	0.7347353980478408	6	None	None
25	discordant	chr12:5317742->chr12:3622528+	3.050039754066433	15	None	None
26	discordant	chr12:5333059+>chr12:2415320-	0.08648889087354909	6	None	None
27	discordant	chr12:5338942+>chr12:1552085-	0.16512269601087262	5	None	None
28	discordant	chr12:5537226+>chr12:4721903+	0.2550143113810229	6	None	None
29	discordant	chr12:5552763+>chr12:1915587-	1.7436357612059463	13	None	None
30	discordant	chr12:5852874->chr12:2416026+	0.7198403408097845	5	None	None
31	discordant	chr12:5930794->chr12:5079556+	0.5128523828624348	7	None	None
32	discordant	chr12:5931375+>chr12:361227+	2.0874044823514742	5	None	None

1	discordant	chr2:111715858->chr12:3704775-	0.3041722201370404	7	None	None
2	discordant	chr2:111916627->chr12:1413208+	1.7017273859991608	8	None	None
3	discordant	chr2:132206280->chr12:4164535-	1.4750332442659695	6	None	None
4	discordant	chr2:132216449->chr12:3365384+	2.031511011492358	11	None	None
5	discordant	chr2:132270851+>chr20:30502688-	0.618978059853053	18	None	None
6	discordant	chr2:132277857+>chr20:30510850-	0.318377052094705	5	None	None
7	discordant	chr2:137926333->chr12:1201557+	0.5587658324337429	3	None	None
8	discordant	chr2:204129577->chr12:1201433-	0.7544055265180644	4	None	None
9	discordant	chr2:204223834->chr12:3524928-	2.348138497728543	7	None	None
10	discordant	chr2:219084633->chr12:4280757-	3.3144486441554677	8	None	None
11	discordant	chr3:15803867+>chr12:3796314-	4.980696978305306	14	None	None
12	discordant	chr3:16400998+>chr12:3366420+	2.0497338155393408	8	None	None
13	discordant	chr3:22039392->chr12:854749+	1.8107170263303538	2	None	None
14	discordant	chr3:35260181->chr12:3856729+	5.31701558211021	12	None	None
15	discordant	chr3:39134453+>chr12:3950650+	2.0005607617192482	8	None	None
16	discordant	chr3:48072033+>chr12:4082889+	2.13934344740282	13	None	None
17	discordant	chr9:2516515->chr10:1366115+	11.422046474167056	34	None	None
18	discordant	chr9:2516515->chr10:1366115+	9.6157850345121	12	None	None
19	discordant	chr9:4096425->chr10:1365625-	6.479996775182927	6	None	None
20	discordant	chr9:4096426->chr10:1365625-	3.4636376066764094	12	None	None
21	discordant	chr9:7033814+>chr10:1365625-	11.33035320296336	33	None	None
22	discordant	chr9:12714213+>chr10:1365941-	6.2000890186782165	76	None	None
23	discordant	chr9:22845823+>chr10:1119733-	14.255067867165161	44	None	None
24	discordant	chr9:80412950+>chr10:61103112-	13.230122569884088	21	None	None
25	discordant	chr9:88731158+>chr10:61102873-	11.477080912337852	17	None	None
26	discordant	chr9:88733484->chr10:7234370+	16.783166954727832	106	None	None
27	discordant	chr9:88734117+>chr10:61103251+	17.666604059879255	59	None	None
28	discordant	chr9:96966842+>chr13:56522571+	22.104092183220402	59	None	None
29	discordant	chr9:101437683->chr10:1366115+	27.639001922752305	111	None	None
30	discordant	chr9:101437862->chr10:1366045-	3.2225637696029095	5	None	None
31	discordant	chr9:116133387->chr13:96876785-	9.910590717151075	50	None	None
32	discordant	chr9:116133635->chr13:56493597-	19.83524760681872	45	None	None
33	discordant	chr9:116133697->chr13:96876797+	5.2889381746585995	8	None	None
34	discordant	chr9:116133757->chr10:61103180+	20.921280431052516	15	None	None
35	discordant	chr9:132728690+>chr10:61103157-	19.13502737598356	26	None	None
36	discordant	chr9:13376415+>chr13:1030540-	10.08730831606530	53	None	None

Part III: Chromosome 3 & 10



Part III: Chromosome 2, 9, 12





Thank you!

Any Questions?