```python
#Setup
import pandas as pd
import numpy as np
import matplotlib.pyplot as plt
import seaborn as sns
import re
from pathlib import Path
```

```python
# Set style
sns.set_style('whitegrid')
plt.rcParams['figure.figsize'] = (12, 6)
```

```python
#Load Data
df = pd.read_csv('texts.csv')
print(f"Loaded {len(df)} texts")
df.head()
```

```
Loaded 60 texts
```

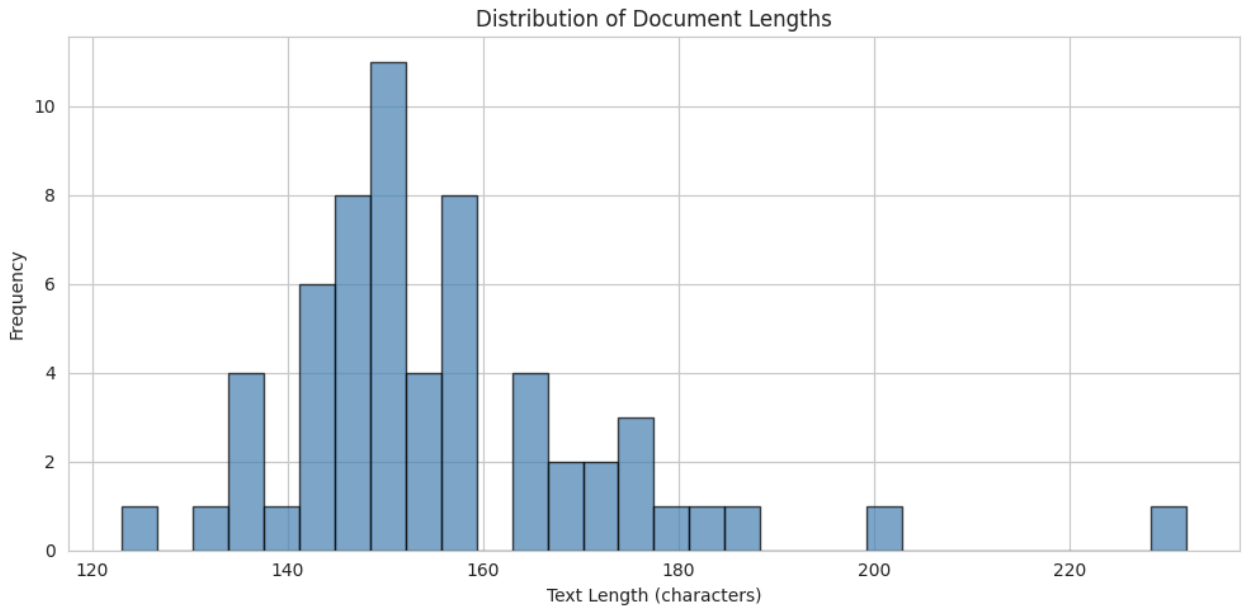
	text_id	title	text
0	T0001	APOE ε4 allele and Alzheimer disease risk	The rs429358 variant in the APOE gene is stron...
1	T0002	TREM2 variants in neurodegeneration	Rare coding variants in TREM2, including rs759...
2	T0003	BIN1 locus and tau pathology	Common variants near BIN1 at rs744373 are asso...
3	T0004	CLU genetic associations in AD	The clusterin gene CLU contains several varian...
4	T0005	CR1 complement receptor variants	Variants in the complement receptor 1 gene CR1...

Next steps: Generate code with df | New interactive sheet

```python
#1. Data Exploration

#Basic Statistics
print(f"Total documents: {len(df)}")
print(f"Average text length: {df['text'].str.len().mean():.0f} characters")
print(f"Unique text IDs: {df['text_id'].nunique()}")
```

```
Total documents: 60
Average text length: 156 characters
Unique text IDs: 60
```

```python
# Text length distribution
plt.figure(figsize=(10, 5))
plt.hist(df['text'].str.len(), bins=30, edgecolor='black', alpha=0.7, color='steelblue')
plt.xlabel('Text Length (characters)')
plt.ylabel('Frequency')
plt.title('Distribution of Document Lengths')
plt.tight_layout()
plt.show()
```




```python
# ## 2. Quick Entity Extraction Test

#Test on Sample
sample_text = df.iloc[0]['text']
print("Sample text:")
print(sample_text)
print("\n" + "="*60 + "\n")
```

```
Sample text:
The rs429358 variant in the APOE gene is strongly associated with increased Alzheimer disease risk across multiple populations. This v
```

```
============================================================
```

```python
# Extract variants
variants = re.findall(r'\brs\d+\b', sample_text)
print(f"Variants found: {variants}")

# Extract gene-like tokens
genes = re.findall(r'\b[A-Z][A-Z0-9]{2,5}\b', sample_text)
genes = [g for g in genes if g not in ['THE', 'AND', 'FOR', 'WITH', 'RNA', 'DNA', 'USA']]
print(f"Potential genes: {genes}")
```

```
Variants found: ['rs429358']
Potential genes: ['APOE']
```

```python
# Look for disease terms
disease_keywords = ['alzheimer', 'dementia', 'disease', 'disorder', 'cognitive', 'parkinson']
found_diseases = [kw for kw in disease_keywords if kw.lower() in sample_text.lower()]
print(f"Disease keywords: {found_diseases}")
```

```
Disease keywords: ['alzheimer', 'disease']
```

```python
# ## 3. Build Entity Extractor

#Entity Extractor Class
class GenomicEntityExtractor:
    def __init__(self):
        self.variant_pattern = re.compile(r'\brs\d+\b')
        self.gene_pattern = re.compile(r'\b[A-Z][A-Z0-9]{2,5}\b')

        self.relations = [
            'associated with', 'linked to', 'increases risk',
            'affects', 'modulates', 'influences', 'causes',
            'contributes to', 'related to', 'implicated in'
        ]

        self.disease_terms = [
            'alzheimer', 'dementia', 'cognitive', 'parkinson',
            'disease', 'disorder', 'syndrome', 'impairment'
        ]

        self.gene_stopwords = {'THE', 'AND', 'FOR', 'WITH', 'RNA', 'DNA', 'USA', 'BUT', 'NOT'}

    def extract_variants(self, text):
        return list(set(self.variant_pattern.findall(text)))

    def extract_genes(self, text):
        candidates = self.gene_pattern.findall(text)
        return list(set([g for g in candidates if g not in self.gene_stopwords]))

    def extract_diseases(self, text):
        diseases = []
        text_lower = text.lower()

        # Find sentences with disease terms
        sentences = text.split('.')
        for sent in sentences:
            sent_lower = sent.lower()
            for term in self.disease_terms:
                if term in sent_lower:
                    # Extract capitalized phrases
                    words = sent.strip().split()
                    for i, word in enumerate(words):
                        if term in word.lower() and len(word) > 2:
                            # Get context window
                            start = max(0, i-2)
                            end = min(len(words), i+3)
                            phrase = ' '.join(words[start:end])
                            if len(phrase) > 5:
                                diseases.append(phrase.strip())

        return list(set(diseases))

    def extract_relations(self, text):
        found = []
        text_lower = text.lower()
        for rel in self.relations:
            if rel in text_lower:
                found.append(rel)
        return found

    def extract_all(self, text, text_id):
        variants = self.extract_variants(text)
        genes = self.extract_genes(text)
        diseases = self.extract_diseases(text)
        relations = self.extract_relations(text)

        records = []

        # Create combinations
```

```
        if variants and genes and diseases and relations:
            for variant in variants:
                for gene in genes:
                    for disease in diseases:
                        for relation in relations:
                            # Find evidence
                            sentences = text.split('.')
                            evidence = ""
                            for sent in sentences:
                                if variant in sent and gene in sent:
                                    evidence = sent.strip()
                                    break

                            records.append({
                                'text_id': text_id,
                                'variant': variant,
                                'gene': gene,
                                'phenotype': disease,
                                'relation': relation,
                                'evidence_span': evidence[:200]
                            })
        else:
            # Create partial records
            if variants or genes or diseases:
                records.append({
                    'text_id': text_id,
                    'variant': variants[0] if variants else None,
                    'gene': genes[0] if genes else None,
                    'phenotype': diseases[0] if diseases else None,
                    'relation': relations[0] if relations else None,
                    'evidence_span': text[:200]
                })

        return records
```

```
# ## 4. Extract Entities from All Texts

# Full Extraction
extractor = GenomicEntityExtractor()

all_entities = []
for idx, row in df.iterrows():
    entities = extractor.extract_all(row['text'], row['text_id'])
    all_entities.extend(entities)

entities_df = pd.DataFrame(all_entities)
print(f"\nExtracted {len(entities_df)} entity records")
print(f"From {len(df)} documents")

# Remove None values
entities_df = entities_df.dropna(subset=['variant', 'gene'])

print(f"After filtering: {len(entities_df)} records")
entities_df.head(10)
```

```
Extracted 97 entity records
From 60 documents
After filtering: 69 records
```

	text_id	variant	gene	phenotype	relation	evidence_span
0	T0001	rs429358	APOE	variant increases disease risk by	associated with	The rs429358 variant in the APOE gene is stron...
1	T0001	rs429358	APOE	with increased Alzheimer disease risk	associated with	The rs429358 variant in the APOE gene is stron...
2	T0001	rs429358	APOE	increased Alzheimer disease risk across	associated with	The rs429358 variant in the APOE gene is stron...
3	T0002	rs75932628	TREM2	late-onset Alzheimer disease	None	Rare coding variants in TREM2, including rs759...
4	T0003	rs744373	BIN1	with Alzheimer disease risk	associated with	Common variants near BIN1 at rs744373 are asso...
5	T0003	rs744373	BIN1	associated with Alzheimer disease risk	associated with	Common variants near BIN1 at rs744373 are asso...
6	T0004	rs11136000	CLU	to Alzheimer disease susceptibility	linked to	The clusterin gene CLU contains several varian...
7	T0004	rs11136000	CLU	to Alzheimer disease susceptibility	influences	The clusterin gene CLU contains several varian...
8	T0004	rs11136000	CLU	linked to Alzheimer disease susceptibility	linked to	The clusterin gene CLU contains several varian...
9	T0004	rs11136000	CLU	linked to Alzheimer disease susceptibility	influences	The clusterin gene CLU contains several varian...

Next steps: Generate code with `entities_df` New interactive sheet

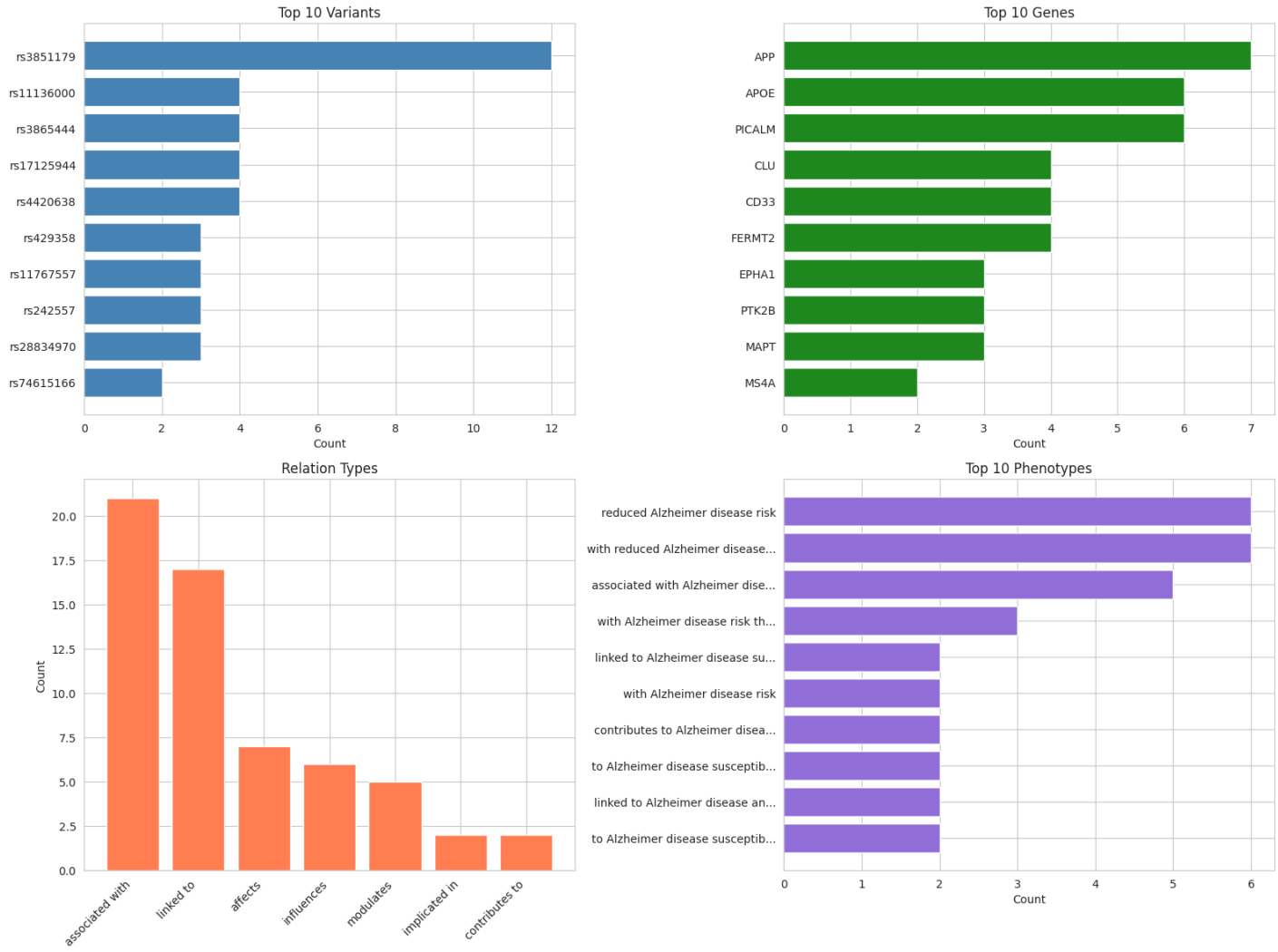
```
# ## 5. Entity Analysis & Visualization

# Entity Statistics
print("="*60)
print("ENTITY STATISTICS")
print("="*60)

print("\nTop 10 Variants:")
print(entities_df['variant'].value_counts().head(10))

print("\nTop 10 Genes:")
```

```
print(entities_df['gene'].value_counts().head(10))

print("\nRelation Types:")
print(entities_df['relation'].value_counts())

print("\nTop Phenotypes:")
print(entities_df['phenotype'].value_counts().head(10))

```

```
============================================================
ENTITY STATISTICS
============================================================

Top 10 Variants:
variant
rs3851179     12
rs11136000     4
rs3865444      4
rs17125944     4
rs4420638      4
rs429358       3
rs11767557     3
rs242557       3
rs28834970     3
rs74615166     2
Name: count, dtype: int64

Top 10 Genes:
gene
APP       7
APOE      6
PICALM    6
CLU       4
CD33      4
FERMT2    4
EPHA1     3
PTK2B     3
MAPT      3
MS4A      2
Name: count, dtype: int64

Relation Types:
relation
associated with    21
linked to          17
affects             7
influences          6
modulates           5
implicated in       2
contributes to      2
Name: count, dtype: int64

Top Phenotypes:
phenotype
reduced Alzheimer disease risk                     6
with reduced Alzheimer disease risk                6
associated with Alzheimer disease risk             5
with Alzheimer disease risk through                3
linked to Alzheimer disease susceptibility         2
with Alzheimer disease risk                        2
contributes to Alzheimer disease susceptibility    2
to Alzheimer disease susceptibility through        2
linked to Alzheimer disease and                    2
to Alzheimer disease susceptibility                2
Name: count, dtype: int64
```

```
#Visualize Entities
fig, axes = plt.subplots(2, 2, figsize=(16, 12))
# Top variants
top_variants = entities_df['variant'].value_counts().head(10)
axes[0, 0].barh(range(len(top_variants)), top_variants.values, color='steelblue')
axes[0, 0].set_yticks(range(len(top_variants)))
axes[0, 0].set_yticklabels(top_variants.index)
axes[0, 0].set_xlabel('Count')
axes[0, 0].set_title('Top 10 Variants')
axes[0, 0].invert_yaxis()

# Top genes
top_genes = entities_df['gene'].value_counts().head(10)
axes[0, 1].barh(range(len(top_genes)), top_genes.values, color='forestgreen')
axes[0, 1].set_yticks(range(len(top_genes)))
axes[0, 1].set_yticklabels(top_genes.index)
axes[0, 1].set_xlabel('Count')
axes[0, 1].set_title('Top 10 Genes')
axes[0, 1].invert_yaxis()

# Relations
relation_counts = entities_df['relation'].value_counts()
axes[1, 0].bar(range(len(relation_counts)), relation_counts.values, color='coral')
axes[1, 0].set_xticks(range(len(relation_counts)))
axes[1, 0].set_xticklabels(relation_counts.index, rotation=45, ha='right')
axes[1, 0].set_ylabel('Count')
axes[1, 0].set_title('Relation Types')

# Top phenotypes
top_phenotypes = entities_df['phenotype'].value_counts().head(10)
axes[1, 1].barh(range(len(top_phenotypes)), top_phenotypes.values, color='mediumpurple')
```

```
axes[1, 1].set_yticks(range(len(top_phenotypes)))
axes[1, 1].set_yticklabels([p[:30] + '...' if len(p) > 30 else p for p in top_phenotypes.index])
axes[1, 1].set_xlabel('Count')
axes[1, 1].set_title('Top 10 Phenotypes')
axes[1, 1].invert_yaxis()

plt.tight_layout()
plt.show()
```




```
# ## 6. Topic Modeling - Find Optimal k

# Elbow Method
from sklearn.feature_extraction.text import TfidfVectorizer
from sklearn.cluster import KMeans
```

```
# Create TF-IDF features
vectorizer = TfidfVectorizer(
    max_features=200,
    stop_words='english',
    ngram_range=(1, 2),
    min_df=2
)
X = vectorizer.fit_transform(df['text'])

print(f"TF-IDF matrix shape: {X.shape}")
```

```
TF-IDF matrix shape: (60, 200)
```

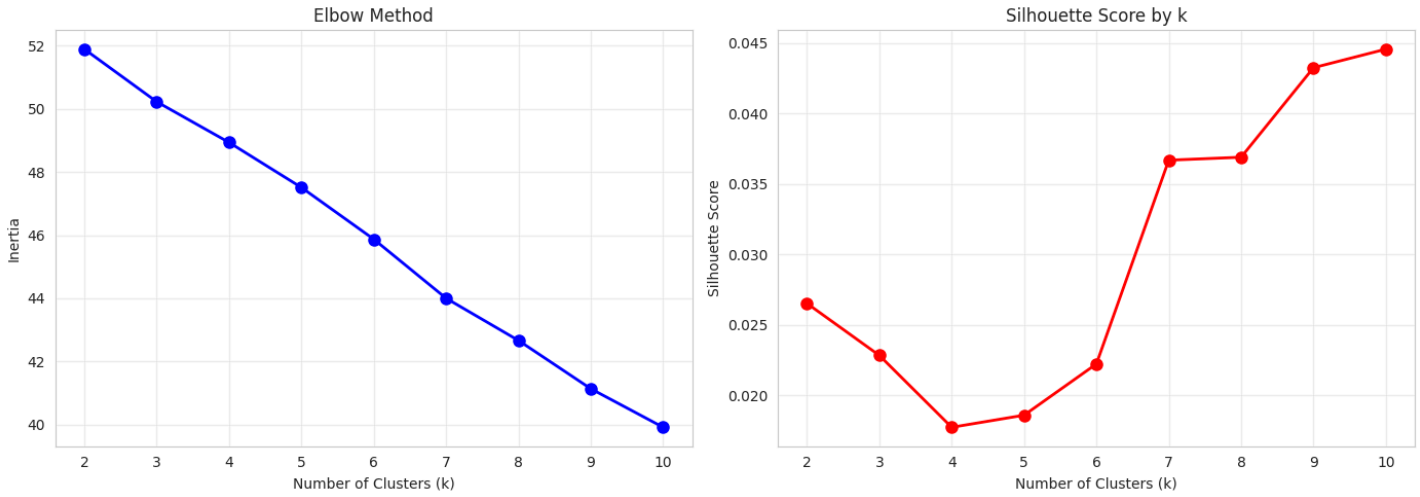
```
from google.colab import drive
drive.mount('/content/drive')
```

```
Drive already mounted at /content/drive; to attempt to forcibly remount, call drive.mount("/content/drive", force_remount=True).
```

```
# Try different k values
inertias = []
```

```
silhouette_scores = []
K_range = range(2, 11)

from sklearn.metrics import silhouette_score

for k in K_range:
    kmeans = KMeans(n_clusters=k, random_state=42, n_init=10)
    labels = kmeans.fit_predict(X)
    inertias.append(kmeans.inertia_)

    # Silhouette score (only if we have enough samples)
    if len(set(labels)) > 1:
        score = silhouette_score(X, labels)
        silhouette_scores.append(score)
    else:
        silhouette_scores.append(0)
```

```
# Plot
fig, (ax1, ax2) = plt.subplots(1, 2, figsize=(14, 5))

ax1.plot(K_range, inertias, 'bo-', linewidth=2, markersize=8)
ax1.set_xlabel('Number of Clusters (k)')
ax1.set_ylabel('Inertia')
ax1.set_title('Elbow Method')
ax1.grid(True, alpha=0.3)

ax2.plot(K_range, silhouette_scores, 'ro-', linewidth=2, markersize=8)
ax2.set_xlabel('Number of Clusters (k)')
ax2.set_ylabel('Silhouette Score')
ax2.set_title('Silhouette Score by k')
ax2.grid(True, alpha=0.3)

plt.tight_layout()
plt.show()

print(f"Best k by silhouette score: {K_range[np.argmax(silhouette_scores)]}")
```



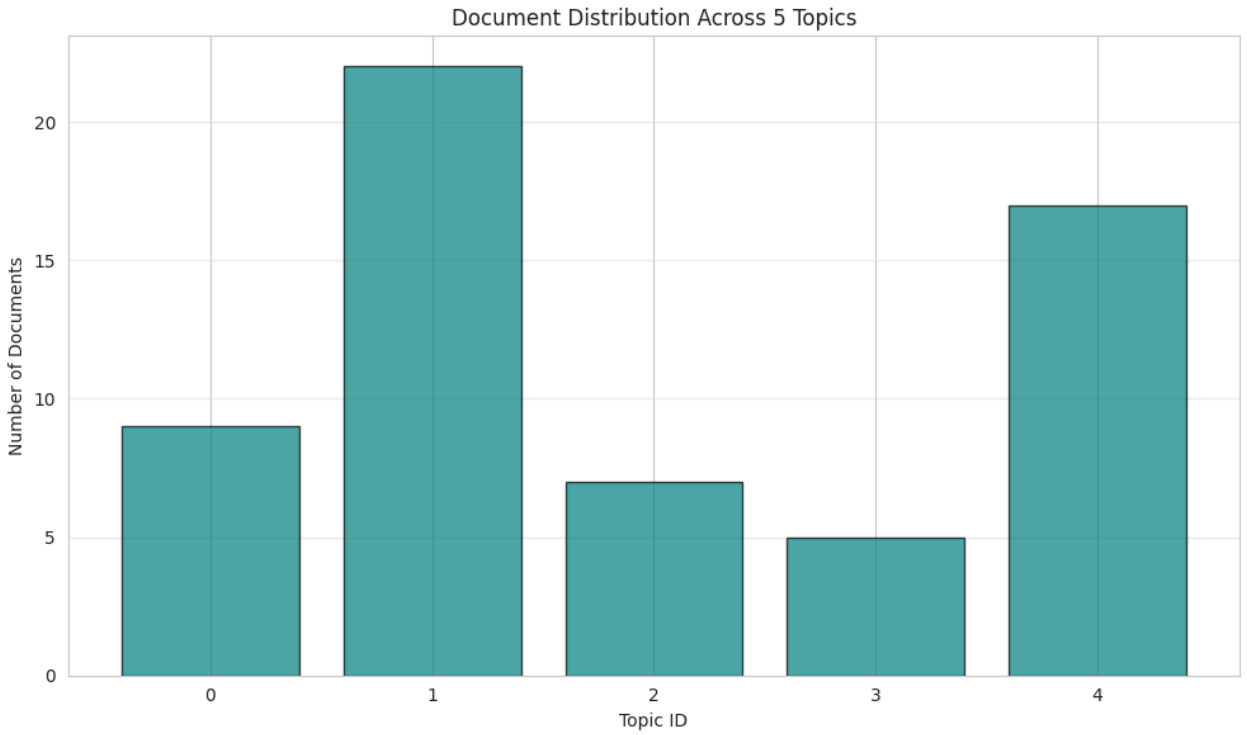

```
Best k by silhouette score: 10
```

```
#7. Final Clustering with k=5

# Cluster Documents
n_clusters = 5
kmeans = KMeans(n_clusters=n_clusters, random_state=42, n_init=20)
df['topic'] = kmeans.fit_predict(X)

# Topic distribution
topic_dist = df['topic'].value_counts().sort_index()
print("\nTopic Distribution:")
print(topic_dist)
```

```
Topic Distribution:
topic
0     9
1    22
2     7
3     5
4    17
Name: count, dtype: int64
```

```
# Visualize distribution
plt.figure(figsize=(10, 6))
plt.bar(topic_dist.index, topic_dist.values, color='teal', alpha=0.7, edgecolor='black')
plt.xlabel('Topic ID')
plt.ylabel('Number of Documents')
```

```
plt.title(f'Document Distribution Across {n_clusters} Topics')
plt.xticks(topic_dist.index)
plt.grid(axis='y', alpha=0.3)
plt.tight_layout()
plt.show()
```




```
# %% Topic Keywords
feature_names = vectorizer.get_feature_names_out()

print("\n" + "="*60)
print("TOPIC KEYWORDS")
print("="*60)

for i in range(n_clusters):
    center = kmeans.cluster_centers_[i]
    top_indices = center.argsort()[-15:][::-1]
    top_words = [feature_names[idx] for idx in top_indices]

    print(f"\nTopic {i} (n={topic_dist[i]} documents):")
    print(f"Keywords: {', '.join(top_words[:10])}")

    # Show 2 example texts
    examples = df[df['topic'] == i].head(2)
    for idx, row in examples.iterrows():
        print(f"  Example: {row['text'][:150]}...")
```

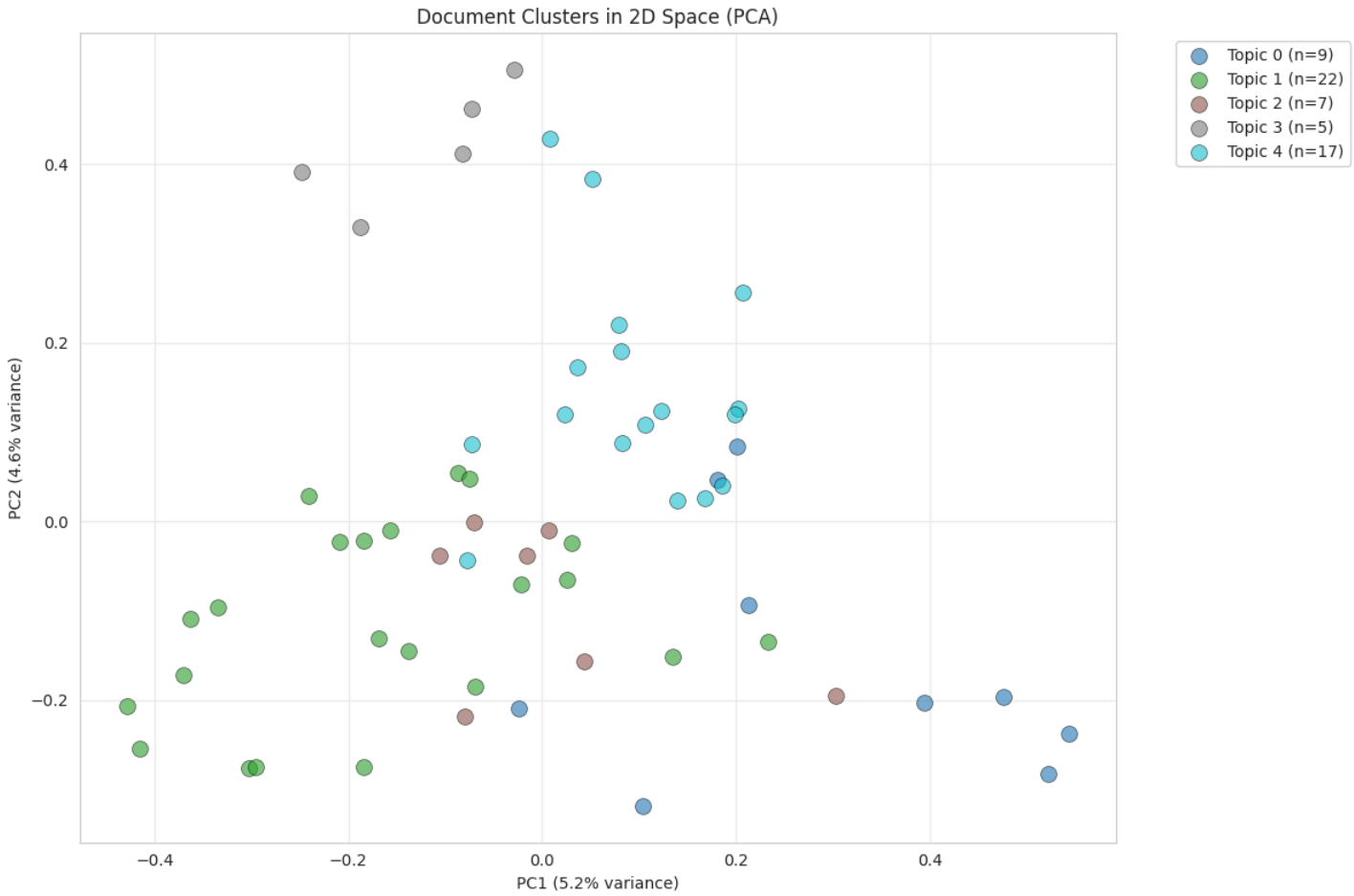
```
============================================================
TOPIC KEYWORDS
============================================================

Topic 0 (n=9 documents):
Keywords: linked, linked alzheimer, synaptic, neuronal, disease, alzheimer disease, alzheimer, variants, cognitive decline, decline
  Example: The clusterin gene CLU contains several variants including rs11136000 that are linked to Alzheimer disease susceptibility.
  Example: The protein tyrosine kinase PTK2B contains variant rs28834970 linked to cognitive decline and Alzheimer disease. PTK2B regu

Topic 1 (n=22 documents):
Keywords: genetic, ad, variants, including, loci, abca7, genetic variants, populations, risk, ad risk
  Example: Loss-of-function variants in ABCA7, including rs3764650, increase AD susceptibility. ABCA7 regulates lipid homeostasis and
  Example: Large-scale genome-wide association study meta-analysis identified 29 risk loci for Alzheimer disease including ZCWPW1, ADA

Topic 2 (n=7 documents):
Keywords: contribute, cd2ap, pathogenesis, blood brain, brain barrier, barrier, blood, brain, genes, contributes
  Example: Variants in the complement receptor 1 gene CR1, particularly rs6656401, contribute to AD risk through altered complement ca
  Example: The immune-related phosphatase gene INPP5D harbors rs35349669 which modulates microglial responses and contributes to AD pa

Topic 3 (n=5 documents):
Keywords: app, beta, amyloid beta, production, beta production, amyloid, psen1, psen2, app psen1, psen1 psen2
  Example: The PICALM gene variant rs3851179 affects clathrin-mediated endocytosis and is associated with reduced Alzheimer disease ri
  Example: Variants in SORL1 such as rs11218343 influence APP trafficking and processing. SORL1 dysfunction leads to increased amyloid

Topic 4 (n=17 documents):
Keywords: risk, disease risk, alzheimer disease, alzheimer, disease, variant, associated, function, gene, multiple
  Example: The rs429358 variant in the APOE gene is strongly associated with increased Alzheimer disease risk across multiple populati
  Example: Rare coding variants in TREM2, including rs75932628, significantly affect microglial function and increase risk for late-or
```

```
# ## 8. Visualize Clusters in 2D (PCA)

#PCA Visualization
from sklearn.decomposition import PCA

# Reduce to 2D
```

```
pca = PCA(n_components=2, random_state=42)
X_2d = pca.fit_transform(X.toarray())

print(f"Explained variance: {pca.explained_variance_ratio_.sum():.2%}")
```

```
Explained variance: 9.88%
```

```
# Plot
plt.figure(figsize=(12, 8))
colors = plt.cm.tab10(np.linspace(0, 1, n_clusters))

for i in range(n_clusters):
    mask = df['topic'] == i
    plt.scatter(
        X_2d[mask, 0],
        X_2d[mask, 1],
        c=[colors[i]],
        label=f'Topic {i} (n={sum(mask)})',
        alpha=0.6,
        s=100,
        edgecolors='black',
        linewidth=0.5
    )

plt.xlabel(f'PC1 ({pca.explained_variance_ratio_[0]:.1%} variance)')
plt.ylabel(f'PC2 ({pca.explained_variance_ratio_[1]:.1%} variance)')
plt.title('Document Clusters in 2D Space (PCA)')
plt.legend(bbox_to_anchor=(1.05, 1), loc='upper left')
plt.grid(True, alpha=0.3)
plt.tight_layout()
plt.show()
```



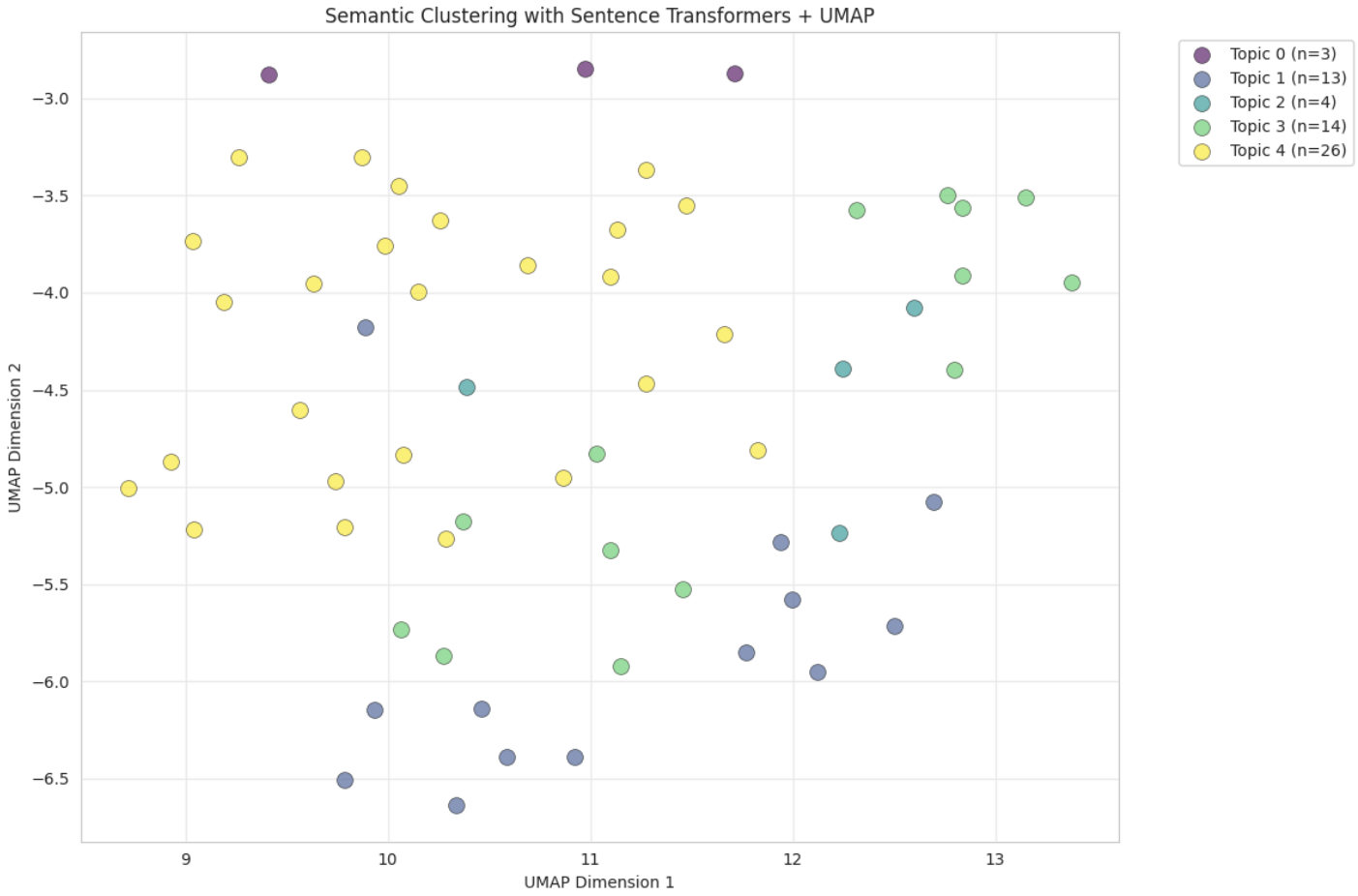

```
# ## 9. Advanced: Semantic Embeddings with UMAP

# Sentence Transformers

try:
    from sentence_transformers import SentenceTransformer
    import umap

    print("Creating semantic embeddings...")
    model = SentenceTransformer('all-MiniLM-L6-v2')
    embeddings = model.encode(df['text'].tolist(), show_progress_bar=True)

    print("Reducing dimensions with UMAP...")
    reducer = umap.UMAP(
        n_components=2,
        random_state=42,
        n_neighbors=15,
```

```
        min_dist=0.1,
        metric='cosine'
    )
    embeddings_2d = reducer.fit_transform(embeddings)

    # Re-cluster on embeddings
    kmeans_emb = KMeans(n_clusters=n_clusters, random_state=42)
    df['topic_semantic'] = kmeans_emb.fit_predict(embeddings)

    # Plot
    plt.figure(figsize=(12, 8))
    colors = plt.cm.viridis(np.linspace(0, 1, n_clusters))

    for i in range(n_clusters):
        mask = df['topic_semantic'] == i
        plt.scatter(
            embeddings_2d[mask, 0],
            embeddings_2d[mask, 1],
            c=[colors[i]],
            label=f'Topic {i} (n={sum(mask)})',
            alpha=0.6,
            s=100,
            edgecolors='black',
            linewidth=0.5
        )

    plt.xlabel('UMAP Dimension 1')
    plt.ylabel('UMAP Dimension 2')
    plt.title('Semantic Clustering with Sentence Transformers + UMAP')
    plt.legend(bbox_to_anchor=(1.05, 1), loc='upper left')
    plt.grid(True, alpha=0.3)
    plt.tight_layout()
    plt.show()

    print("\nSemantic clustering complete!")

except ImportError:
    print("\nSkipping semantic embeddings (sentence-transformers not installed)")
    print("To enable: pip install sentence-transformers umap-learn")

```

```
Creating semantic embeddings...
/usr/local/lib/python3.12/dist-packages/huggingface_hub/utils/_auth.py:94: UserWarning:
The secret `HF_TOKEN` does not exist in your Colab secrets.
To authenticate with the Hugging Face Hub, create a token in your settings tab (https://huggingface.co/settings/tokens), set it as s
You will be able to reuse this secret in all of your notebooks.
Please note that authentication is recommended but still optional to access public models or datasets.
  warnings.warn(
```

Batches: 100% 2/2 [00:02<00:00, 1.36s/it]

```
Reducing dimensions with UMAP...
/usr/local/lib/python3.12/dist-packages/umap/umap_.py:1952: UserWarning: n_jobs value 1 overridden to 1 by setting random_state. Use
  warn(
```




```
Semantic clustering complete!
```

```python
# ## 10. Save Results

# Export Data
# Create output directory
Path('../output').mkdir(exist_ok=True)

# Save entities
entities_df.to_csv('../output/entities.csv', index=False)
entities_df.to_json('../output/entities.json', orient='records', indent=2)

# Save clustered documents
df.to_csv('../output/texts_with_topics.csv', index=False)

# Create topic summary
topic_summary = []
for i in range(n_clusters):
    center = kmeans.cluster_centers_[i]
    top_indices = center.argsort()[-10:][::-1]
    top_words = [feature_names[idx] for idx in top_indices]

    topic_summary.append({
        'topic_id': i,
        'size': int(topic_dist[i]),
        'keywords': top_words,
        'example_texts': df[df['topic'] == i]['text'].head(3).tolist()
    })

import json
with open('../output/topics.json', 'w') as f:
    json.dump(topic_summary, f, indent=2)

print("\n" + "="*60)
print("RESULTS SAVED")
print("="*60)
print("✓ entities.csv")
```