



Centromere Stability and Chromosome Segregation



Prisha Chatterjee • Crossroads North Middle School • South Brunswick, New Jersey | learningprisha@gmail.com

Mentor: Mariah Antopia

TITLE: Centromere Stability and Chromosome Segregation: Implications for Trisomies, Cancer & Other Genetic Disorders

Introduction: Why Centromeres Matter

- Chromosome segregation is an important process for maintaining genetic stability
- Chromosomes must be copied and separated accurately during cell division.
- The centromere is the chromosome region where the kinetochore forms.
- CENP-A helps mark the centromere and supports kinetochore assembly.
- Spindle fibers attach to kinetochores and pull chromosomes apart.
- If these steps fail, cells can receive the wrong number of chromosomes.

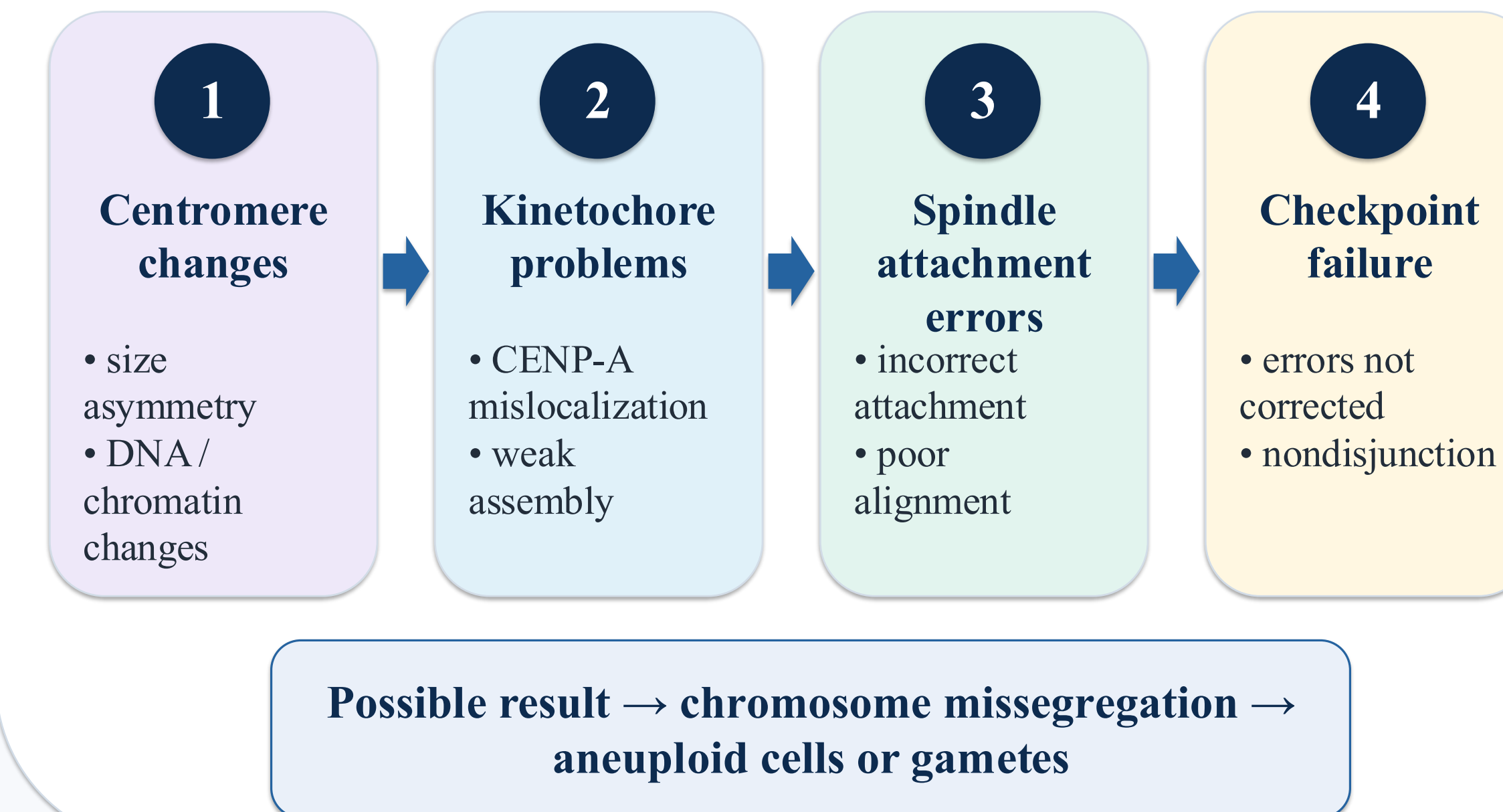
Methods

- Systematic literature review using scientific articles and medical sources.
- Databases included PubMed, Google Scholar, Nature Reviews, and Cold Spring Harbor Perspectives in Biology.
- Methods reviewed included long-read sequencing, epigenomic profiling, transcriptomics, and artificial chromosome systems.

Complexity is same Type of Error, but Different Outcomes

Condition	Extra chromosome	Examples of effects	Overall outcome
Trisomy 13 (Patau)	13	Cleft lip/palate, brain & heart defects	Usually severe; reduced survival
Trisomy 18 (Edwards)	18	Growth restriction, organ abnormalities	Usually severe; reduced survival
Trisomy 21 (Down)	21	Developmental differences, heart/hearing/vision issues	Higher survival than Trisomy 13 or 18

What Can Go Wrong During Chromosome Segregation?



Main Findings : Results

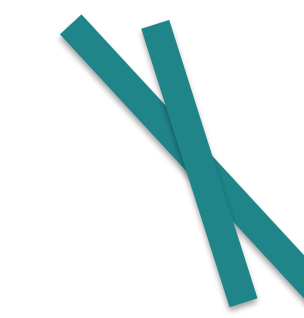
- Centromere stability depends on DNA structure, epigenetic regulation, CENP-A, kinetochore assembly, and spindle-checkpoint control.
- Centromere size asymmetry and methylation differences have been studied in relation to Trisomy 21.
- No current clinical treatment can correct trisomy by fixing chromosome segregation at the chromosomal level.

Why This Matters

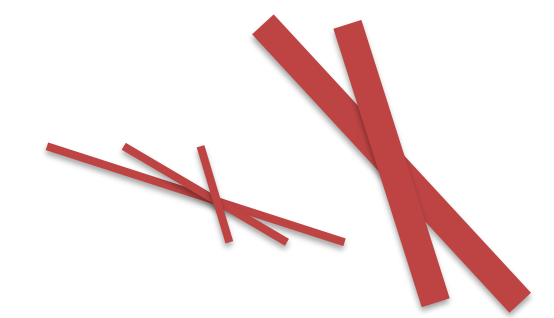
- Studying centromeres can help scientists better understand why chromosome-number errors happen.
- This knowledge may support future research in diagnosis, risk assessment, genetic counseling, cancer biomarkers, and experimental chromosome-engineering approaches.

Connection to Cancer

Stable segregation



Chromosomal instability



Cancer cells often show abnormal chromosome numbers and chromosome instability. Altered centromere/kinetochore function and CENP-A expression are being studied as possible biomarkers.

Discussion

- Chromosome missegregation is not caused by one factor; several centromere and cell-division systems work together.
- Trisomies 13, 18, and 21 share a nondisjunction mechanism but have different effects.
- The same broad topic—chromosome instability—is also important in cancer biology and other genetic disorder
- Many future therapies discussed in the literature are still experimental.
- Efficacies of drugs like Shugoshin-1 reduce age related chromosomal abnormalities

Conclusion

- The centromere is a small chromosome region with a very important job. Its DNA, epigenetic state, CENP-A, kinetochore, spindle attachments, and checkpoint proteins must work together for accurate chromosome separation.
- When these systems fail, chromosome-number errors can occur.
- Understanding these mechanisms can improve research, therapeutics and management of trisomies, cancer, and other chromosome-related issues, even though these remain largely experimental with no preventive or corrective clinical intervention.

SELECTED REFERENCES

Gorbsky (2016) • Krivega et al. (2022) • Jaggi et al. (2026) • Jiang et al. (2026) • Kumara & Mastrosa (2021) • Hogan et al. (2026) • Achrem (2020) • Bakhoun et al. (2015) • Yildirim (2026) • MedlinePlus (2022)