

Mitochondrial cytopathy is an older, umbrella term used to describe a heterogeneous group of disorders resulting from **mitochondrial dysfunction**, specifically involving defects in the **mitochondrial respiratory chain**, leading to impaired **ATP production**.

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□ What does the term mean?

* **“Cytopathy”** refers to cellular damage or disease; in this case, due to mitochondrial impairment. * It encompasses disorders caused by mutations in either **mitochondrial DNA (mtDNA)** or **nuclear DNA** that affect proteins essential for **oxidative phosphorylation**. * The clinical spectrum is broad, ranging from **isolated organ involvement** (e.g., optic atrophy or myopathy) to **severe multisystem syndromes** like **MELAS**, **Leigh syndrome**, or **MERRF**.

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□ Why is the term considered outdated or imprecise?

* It does **not specify the genetic cause** or the exact molecular defect. * It fails to indicate **which respiratory chain complex** is affected. * It blurs the line between **primary mitochondrial disorders** (genetically defined) and **secondary mitochondrial dysfunction** (e.g., due to toxins, aging, or acquired disease). * Modern classifications prefer terms like **“primary mitochondrial disease (PMD)”**, which reflect the **genotype-phenotype relationship** and guide diagnostic and therapeutic strategies more precisely.

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□ Summary

* **“Mitochondrial cytopathy”** = a broad, historical term. * Now largely replaced by more specific nomenclature. * Still used in some clinical contexts when the diagnosis is **suspected but not genetically confirmed**.

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