Flex Therapist CEUs

Rett Syndrome: Biology, Development, and Prognosis

Rett syndrome - biological pathways leading from MECP2 to disorder phenotypes

1. All MECP2 mutations cause RTT and all RTT patients have mutated MECP2.

A. True
B. False

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2. MECP2 affects epigenetic regulation of gene expression, which changes _____, causing the major phenotype.

A. Neurobiological activity
B. Network formation
C. Network function
D. All of the above

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3. All of the following are the most abundant causes of death of RTT females, except for:

A. Upper respiratory tract infection
B. Lower respiratory tract infection
C. Aspiration / asphyxication
D. Respiratory failure

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4. MECP2 protein is most abundant in which tissue?

A. Lung
B. Brain
C. Spleen
D. Muscle

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5. The main function of MECP2 is to recognize and bind specifically methylated cytosine residues in the DNA that are enriched with A/T bases adjacent.

A. True
B. False

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6. The molecular functions of MECP2 are known to influence which biological mechanism?

A. MECP2 influences global translation by enhancing the AKT/mTOR signaling pathway.
B. Alternative splicing of downstream gene products is affected because MECP2 forms a complex with YB1, an important splicing factor.
C. MECP2 triggers the chromatin compaction at methylated DNA sites which regulates the transcription of adjacent genes.
D. All of the above.

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7. Most of the mutations which cause RTT occur in the transcriptional repression domain (TRD) region of MECP2.

A. True
B. False

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8. Early autopsies revealed reduced levels of catecholamines, namely _____.

A. Dopamine
B. Serotonin
C. Norepinephrine
D. All of the above

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9. Glutamate has been shown to be present in high levels in RTT females, which may lead to over excitation of glutamatergic neurons and can trigger increased uptake and conversion of glutamate to glutamine.

A. True
B. False

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10. In the absence of functional MECP2, the expression of _____, a MECP2 regulated protein necessary for neuronal development and function, is upregulated.

A. GAMT
B. FKBP5
C. BDNF
D. All of the above

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11. Activation of a single pathway seems to contribute more to disorder development than the balance and timing of transcription levels.

A. True
B. False

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12. MECP2 mutations cause RTT by disrupting which major function?

A. Co-repressor recruitment
B. Chromatin compaction
C. Both (A) and (B)
D. None of the above

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13. In a premature state, MECP2 acts as an activator of transcription, while transcription repressor activity is only found in mature neurons.

A. True
B. False

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14. An abnormal ratio of excitation / inhibition in brain activity has also been found in:

A. Autistic patients
B. Parkinson's disease
C. Both (A) and (B)
D. None of the above

Developmental delay in Rett syndrome: data from the natural history study

15. When comparing motor behavioral assessments, which of the following were found to be better among individuals who attained better scores?

A. Gross motor skills and receptive communication
B. Fine motor skills and receptive communication
C. Gross motor skills and expressive communication
D. Fine motor skills and expressive communication

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16. Individuals with R133C, R294X, R306C, and 3' truncations may have a greater amount of all of the following behavioral issues, except for:

A. Anxiety
B. Frequent / severe tantrums
C. Aggressiveness
D. Inappropriate activities

Twenty years of surveillance in Rett syndrome: what does this tell us?

17. Which of the following is a common comorbidity of Rett syndrome?

A. Gastrointestinal problems
B. Sleep disturbance
C. Epilepsy
D. All of the above

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18. Gross motor capabilities are influenced by the type of mutation present in the MECP2 gene.

A. True
B. False

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19. A longitudinal study of adult women with Rett syndrome indicated that age-related deterioration of gross motor function is fast.

A. True
B. False

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20. Loss of walking is inevitable as women with Rett syndrome age.

A. True
B. False
