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November 13-14th 2018

Mercure Hotel Padang, West Sumatra

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ASSOCIATION BETWEEN THE SNP rs2154545 POLYMORPHISM ON THE DyrkIA GENE AND INTELLECTUAL DISABILITY IN DOWN SYNDROME

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INTRODUCTION

- DS caused by meiotic non-disjunction, Robertsonian translocation, mosaic non-disjunction & rearrangement of the genetic material on chromosome 21 with other acrocentric chromosomes.
- Etiology & pathology → focused on extra copy of the region in the proximal position of chromosome 21 (21q22).

INTRODUCTION

- Down syndrome (DS) —a major pediatric issue due to its impact on growth & development
- A genetic abnormality (intellectual disability, hypotonia, facial dysmorphic, earlier onset of Alzheimer's disease & other behavior disruptions)

INTRODUCTION

- Pathogenesis → phenotype abnormalities, (morphology figure, hypotonia & intellectual disability).
- Latest research → DyrkIA gene located at 21q22.3 → long-arm of chromosome 21.
- This location is 5.4 Mb in size and contains 30 genes, based on the phenotype & genotype correlation in a partial trisomy 21 case study.

INTRODUCTION

- The incidence :
- US is estimated to occur once in every 800–1000 birth
- Dr. M. Djamil Hospital Padang →
  - ❖95 DS cases 2009 - 2012,
  - ❖112 DS cases from 2013 -2016

INTRODUCTION

- DyrkIA (overexpressed in DS) : the center of the multiple pathway deregulations in brain development & aging with structural and functional damage.
- DrykIA overexpression → early-onset neurofibril degeneration by hyperphosphorylation of the tau protein & indirectly by the phosphorylation of alternative splicing factor (ASF),which imbalances the 3R-tau & 4R-tau proteins

CONCLUSION

- The results: there are an observable SNP rs2154545 polymorphism on the DrykIA gene has heterozygote mutant (Allele GA)
- Causes the most common clinical manifestation of this overexpression is a mild–borderline intellectual disability in people with DS.

THANK YOU