# CURRICULUM VITAE

## Dr. dr. Dovy Djanas, Sp.OG (K-FM)

- Tempat / Tgl lahir : Padang / 19 April 1968
- S1 : Kedokteran Universitas Andalas.
- Spesialis 1 : Obgin FK Unand-RSUP Dr.M. Djamil Padang, 2005
- Spesialis 2 : Fetomaternal FKUI-RSCM Jakarta, 2011
- S 3 : Biomedik FK Unand, 2018
- Pendidikan S2 MARS
- Klinisi di Klinik Morula IVF RS Bunda BMC Padang
- Staff Obgin FK-Unand / RSUP Dr. Djamil Padang
- Kepala Instalasi Rawat Inap Ibu dan Anak RSUP Dr. M. Djamil Padang
- Ketua POGI Sumbar 2019 - 2021
- Pemakalah Tetap Media Koran Padang Ekspress 2012 sampai sekarang

# DETEKSI DINI CACAT LAHIR PADA ANC

DOVY DJANAS

DIVISI FETOMATERNAL BAGIAN OBSTETRI DAN GINEKOLOGI
FK UNAND/ RS INTERNASIONAL DR M DJAMIL PADANG

# PENDAHULUAN

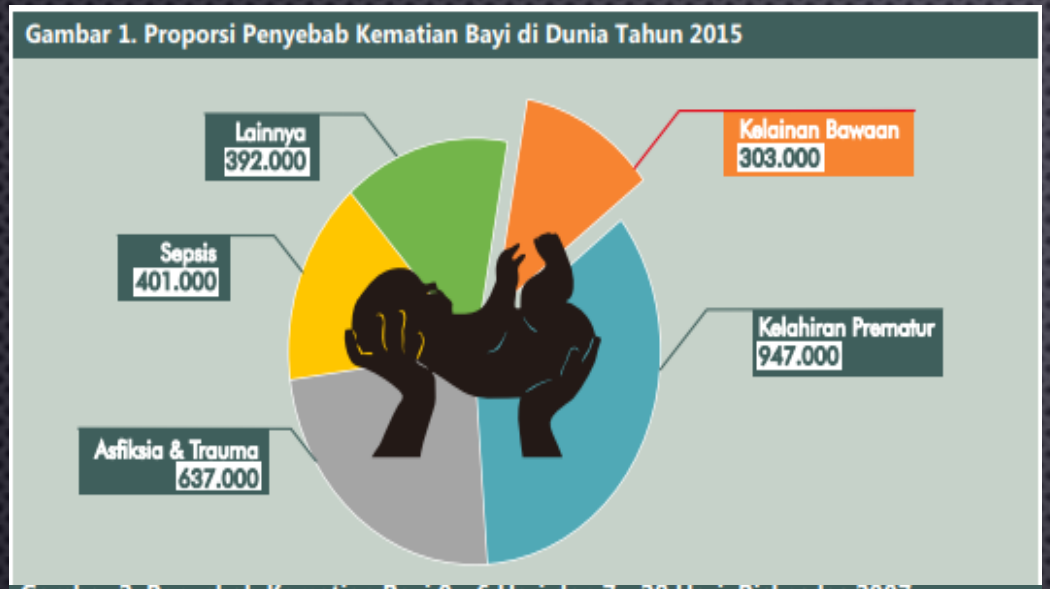
Lebih dari 4 juta anak dilahirkan dengan cacat lahir setiap tahun

**Merupakan tantangan kesehatan klinis dan publik yang cukup besar karena dampaknya di seluruh dunia terhadap kesehatan populasi**

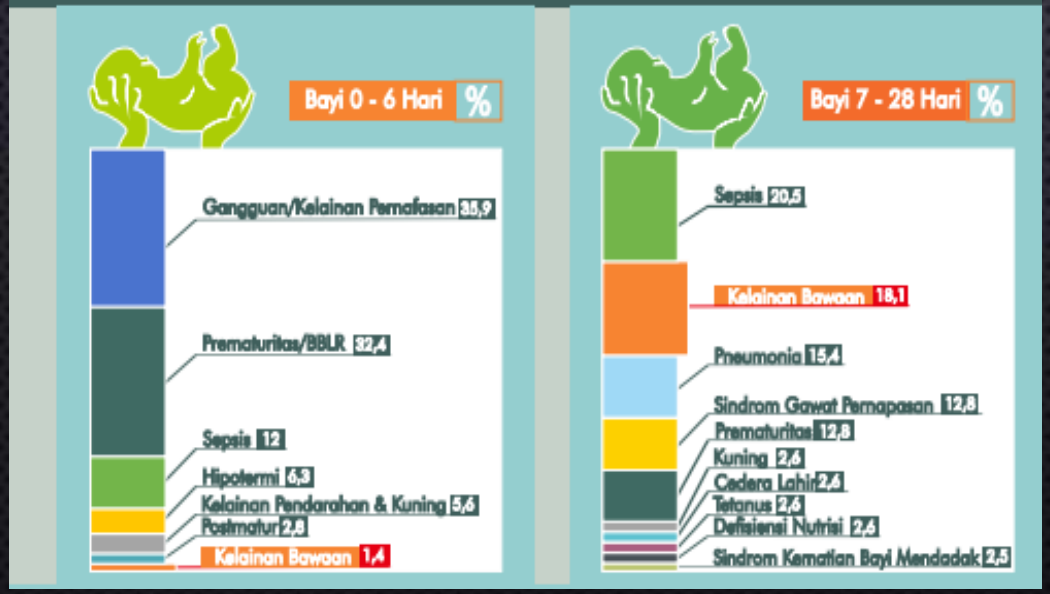
*WHO :*
*Dari 2,68 juta kematian bayi, 11,3% disebabkan oleh kelainan bawaan (INFODATIN Kemenkes RI, 2014-2018)*

Di Indonesia, hasil Riskesdas tahun 2007 menjelaskan kelainan bawaan menjadi salah satu penyebab kematian bayi

Gambar 1. Proporsi Penyebab Kematian Bayi di Dunia Tahun 2015




Gambar 2. Penyebab Kematian Bayi 0 - 6 Hari dan 7 - 28 Hari, Riskesdas 2007

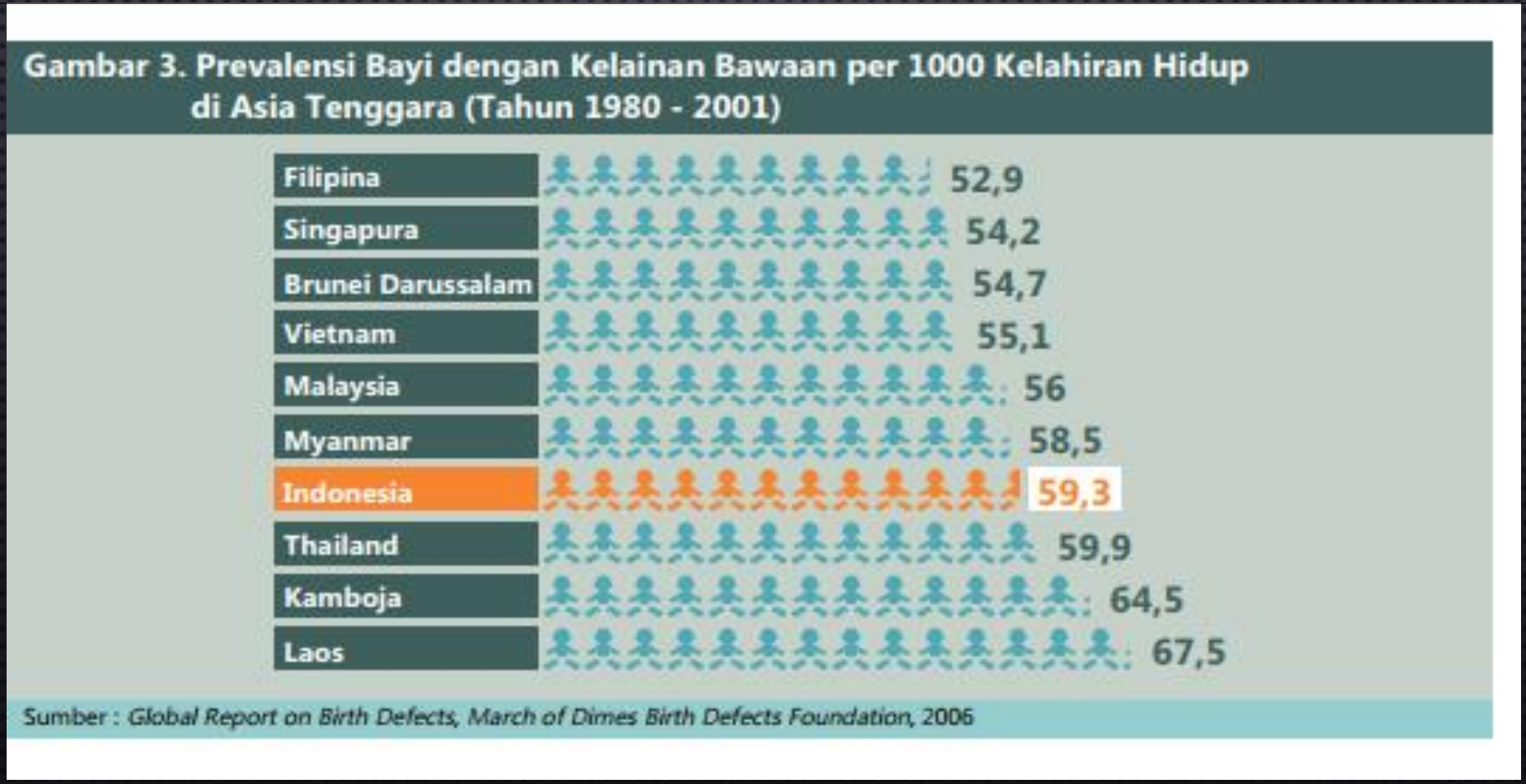



| Order | Birth defects | Cases | Prevalence(1/10000) |
|---|---|---|---|
| 1 | Neural tube defects | 126 | 20.1 |
| 2 | Congenital heart disease | 107 | 17.1 |
| 3 | Cleft lip and palate | 101 | 16.2 |
| 4 | Congenital hydrocephalus | 62 | 9.9 |
| 5 | Polydactylia | 53 | 8.5 |
| 6 | External ear malformation | 27 | 4.3 |
| 7 | Limb reduction defect | 26 | 4.2 |
| 8 | Inguinal hernia | 25 | 4.0 |
| 9 | Congenital hemangiomas | 18 | 2.9 |
| 10 | Talipes equinovarus | 17 | 2.7 |

| Organs or systems | Cases | Proportion(%) |
|---|---|---|
| Central nervous system | 193 | 19.8 |
| Face or eye | 176 | 18.0 |
| Extremities | 129 | 13.2 |
| Cardiovascular | 118 | 12.1 |
| Skeletal | 76 | 7.8 |
| Genitourinary tract | 60 | 6.1 |
| Thoracic or abdominal wall | 55 | 5.7 |
| Gastrointestinal tract | 41 | 4.2 |
| Tumors | 10 | 1.0 |
| Other | 118 | 12.1 |
| Total | 976 | 100.0 |

## SITUASI NASIONAL

*March of Dimes Birth Defects Foundation* pada tahun 2006 merilis *Global Report on Birth Defect* prevalensi bayi dengan kelainan bawaan di Indonesia adalah 59,3 per 1.000 kelahiran hidup



# CACAT LAHIR

NBBD
Newborn Birth Defects





**Suatu kondisi pada saat bayi lahir yang dilihat tepat ketika bayi lahir**

**Kelainan struktur pada janin yang terjadi ketika janin masih berkembang didalam perut ibu**

## Cacat Lahir Struktural : Mayor atau Minor

## Cacat lahir INTERNAL / EKSTERNAL

**Sistem saraf atau masalah otak, masalah sensorik, gangguan metabolisme, gangguan degeneratif**

# APA PENYEBABNYA???

- Masalah genetik
- Masalah kromosom
- Infeksi
- Terpapar obat, bahan kimia, atau agen lain selama kehamilan
- **Gangguan Makronutrien dan Mikronutrien selama kehamilan**

Defisiensi VIT A

Defisiensi Asam Folat

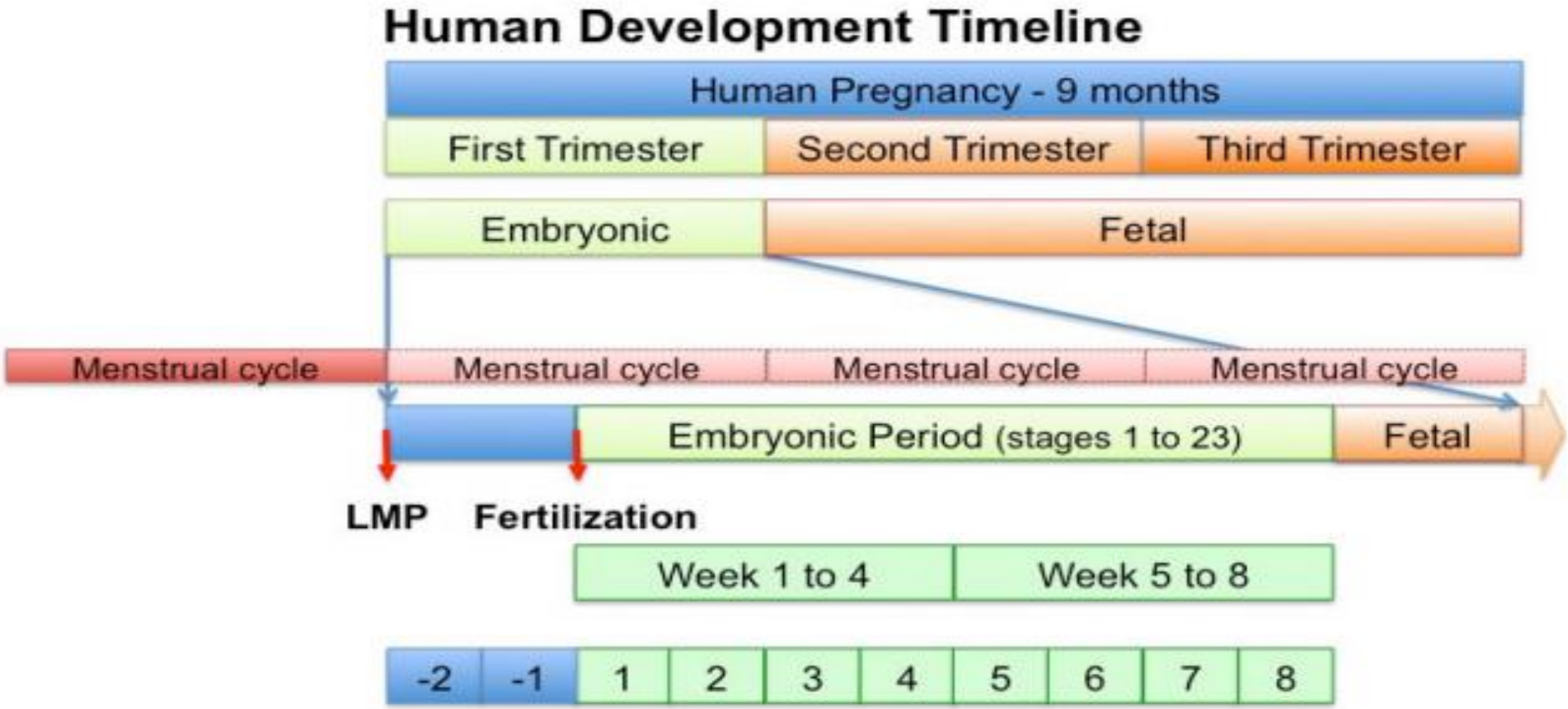
Defisiensi Zn

Defisiensi vitamin E

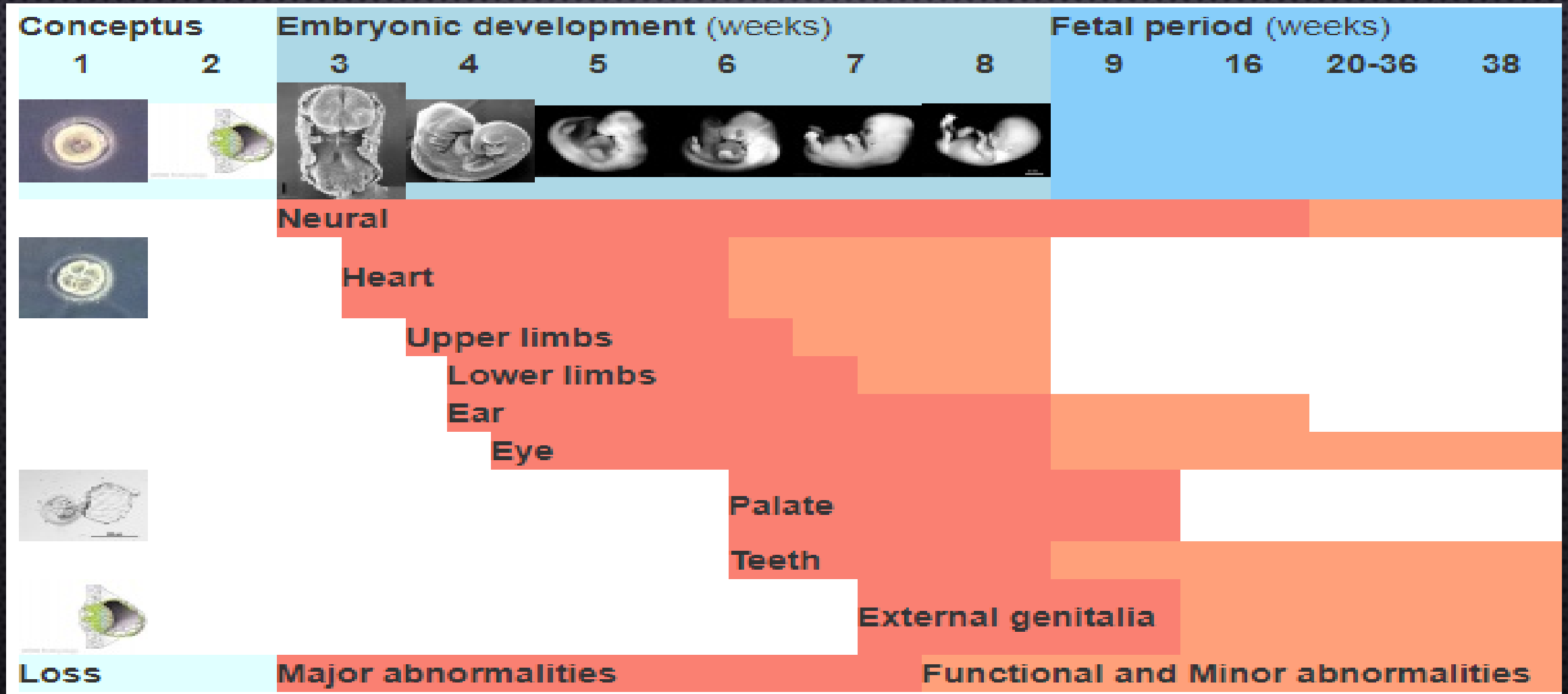
# TABEL KEBUTUHAN ASUPAN MAKRONUTRIEN DAN MIKRONUTRIEN PADA IBU HAMIL DAN TIDAK HAMIL

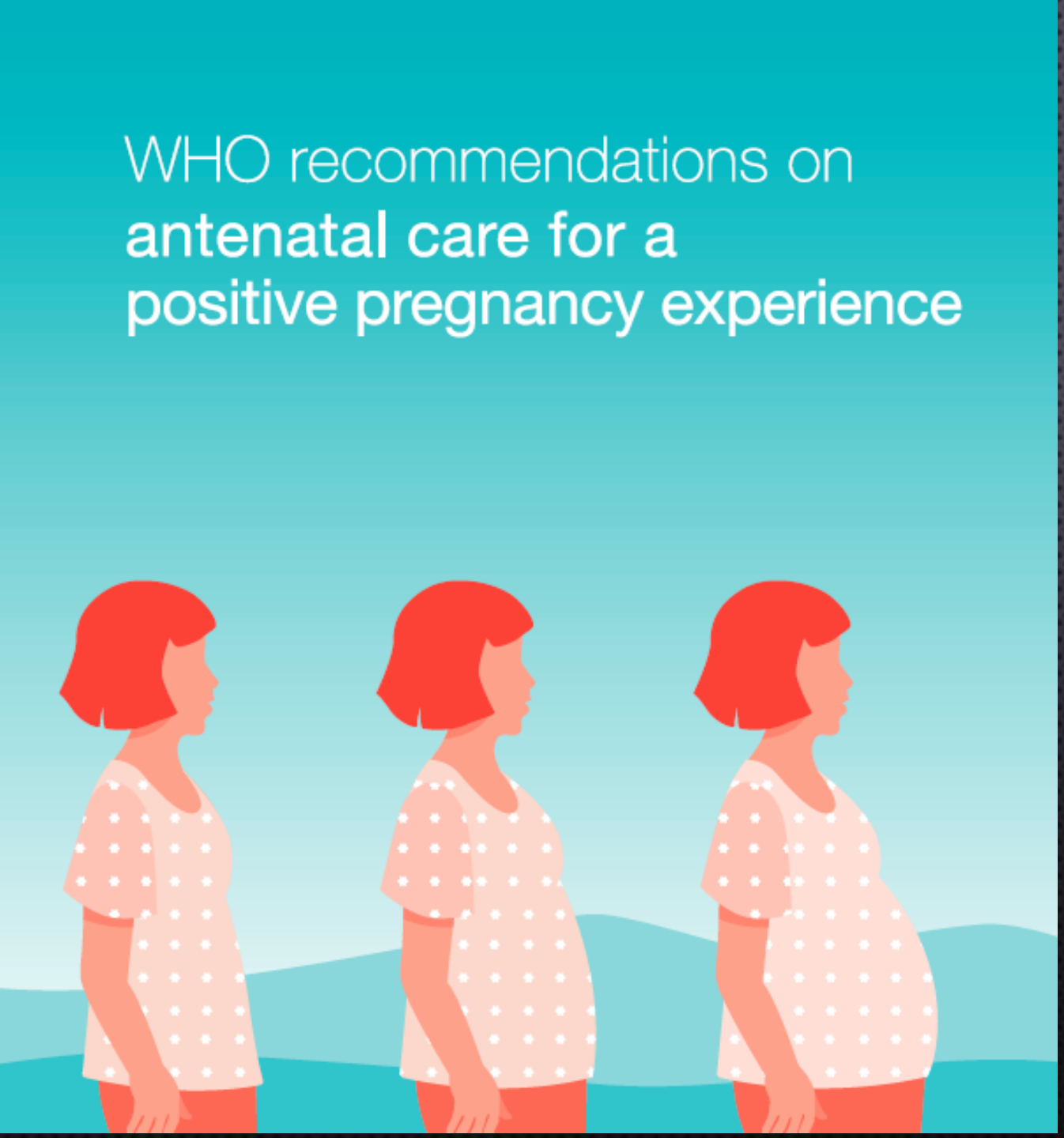
| Nutrient | Recommendations for Interventions/Supplement Use [1] | Non Pregnant Adult Females (19–50 years) [2] | Pregnant Adult Females (19–50 years) [2] |
|---|---|---|---|
| **Macronutrients** | | | |
| Energy | Energy restriction reduces GWG but could adversely affect birthweight and is currently not recommended in pregnancy | EER (kcal/day) [3] = 354 − (6.91 × age [year]) + PA × [(9.36 × weight [kg]) + (726 × height [m])] | Non pregnant EER + 340 and 452 kcal/day in 2nd and 3rd trimesters |
| Protein | Balanced energy/protein supplements (≤ 25% total energy from protein) are recommended only in undernourished women to prevent stillbirth and SGA | 0.8 g/kg/day (46 g/day) | 0.8 increasing to 1.1 g/kg/day in 2nd half of pregnancy (71 g/day) |
| Total fibre [4] | Fibre-rich diet may reduce preeclampsia and GDM but no specific recommendations are currently available; fibre supplements can be used to relieve constipation if diet modification is unsuccessful | 14 g/1000 kcal (or 25 g/day) | 14 g/1000 kcal or (or ~28 g/day to account for GWG) |
| Carbohydrates (GI and GL) | Low GL or GI diets may be beneficial for women at risk of GDM or LGA but can increase risk of SGA. No specific recommendations are currently available | 130 g/day of carbohydrates | 175 g/day of carbohydrates |
| Essential fatty acids [4] (linoleic acid [*n*–6] and α-linoleic acid [*n*–3]) | *n*–3 PUFAs may prevent preterm birth but can increase post-term birth and LGA. No specific recommendations are currently available | 12 g/day (linoleic) 1.1 g/day (α-linoleic) | 13 g/day (linoleic) 1.4 g/day (α-linoleic) |
| **Micronutrients** | | | |
| Folate/folic acid | Recommended (400 µg/day) from preconception until at least 12 weeks to prevent NTDs | 400 µg/day | 600 µg/day |
| Vitamin A | Not recommended except in areas with severe deficiency/night blindness | 700 µg/day | 770 µg/day |
| Thiamine ($B_1$)<br>Niacin ($B_2$)<br>Riboflavin ($B_3$)<br>Pyridoxine ($B_6$)<br>Cyanocobalamin ($B_{12}$) | B-complex vitamins are not recommended to improve pregnancy outcomes until further evidence is available | 1.1 mg/day<br>14 mg/day<br>1.1 mg/day<br>1.3 mg/day<br>2.4 µg/day | 1.4 mg/day<br>18 mg/day<br>1.4 mg/day<br>1.9 mg/day<br>2.6 µg/day |
| Vitamin C<br>Vitamin E | Not recommended until further evidence relating to safety and PROM is available | 75 mg/day<br>15 mg/day | 85 mg/day<br>15 mg/day |
| Vitamin D [4] | Not recommended for improving pregnancy outcomes but should be given to women with deficiency (200 IU/day) | 5 µg/day | 5 µg/day |
| Calcium [4] | Recommended (1.5–2.0 g/day) to prevent hypertensive disorders in women with low dietary calcium intake or who are at high risk of hypertension | 1 g/day | 1 g/day |
| Iodine | Recommended only in women at high risk to prevent IDDs (i.e., in countries where < 20% of households have access to iodized salt) | 150 µg/day | 220–250 µg/day |
| Iron | Recommended (30–60 mg/day) to prevent maternal anaemia, puerperal sepsis, LBW and preterm birth | 18 mg/day | 27–60 mg/day |
| Zinc | Not recommended for improving pregnancy outcomes until more rigorous research is available | 8 mg/day | 11 mg/day |
| Alcohol | Not recommended during pregnancy until safe upper limits are established | NA | None |
| Caffeine | Reducing intake is recommended in women with high caffeine intake (> 300 mg/day) to prevent pregnancy loss and LBW infants | NA | <200 mg/day |

# TIMELINE KEHAMILAN



# Human Citical Periods of Development






# WHO GUIDELINES

A. Intervensi nutrisi
B. **Penilaian ibu dan janin**
C. **Tindakan pencegahan**
D. Intervensi untuk gejala fisiologis umum
E. Intervensi sistem kesehatan untuk meningkatkan pemanfaatan dan kualitas ANC.

# SKRINING CACAT LAHIR KETIKA ANTENATALCARE

## TRIMESTER 1 pada 9–13 MINGGU KEHAMILAN

## TRIMESTER 2 pada 14-18 MINGGU KEHAMILAN

- Tes darah sederhana (Serum ibu)
- Ultrasonografi Anomali

# HASILNYA?

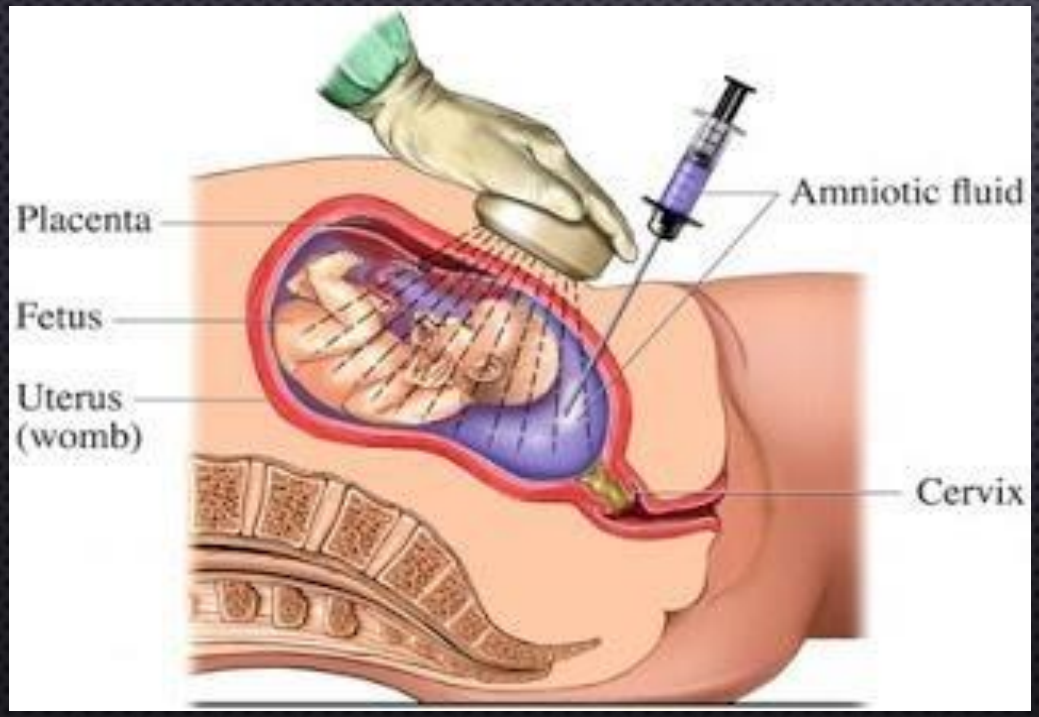
INCREASED RISK

NOT INCREASED RISK

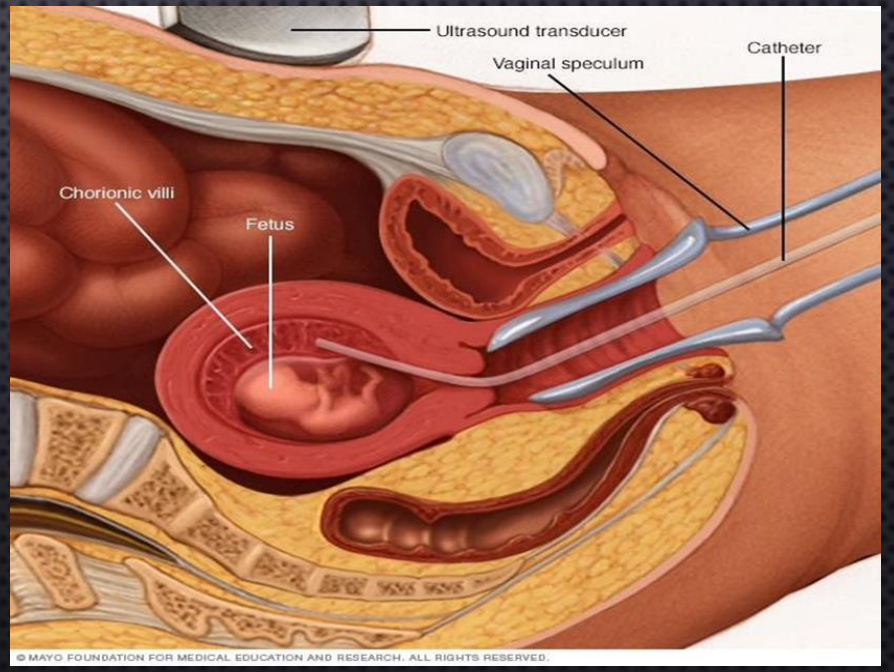
# TES DIAGNOSTIK

1. **Tes  prenatal noninvasif (NIPT)**
2. **Amniosentesis**
3. **Chorionic Villus Sampling (CVS)**
4. Ultrasound resolusi tinggi
5. Alpha Fetoprotein (AFP
6. Acetylcholinesterase (AChE)

# Amniosentesis




# Chorionic Villus Sampling (CVS)



# APA SAJA TINDAKAN PREVENTIF???

# INTERVENSI GIZI

- Intervensi diet
- Suplemen zat besi dan asam folat
- Suplemen kalsium
- Suplemen vitamin A
- Suplemen seng
- Suplemen Multiple Micronutrients (MMN)
- Suplemen vitamin B6 (pyridoxine)
- Suplemen vitamin E & C
- Suplemen vitamin D
- Membatasi asupan kafein

*Review*

# Macronutrient and Micronutrient Intake during Pregnancy: An Overview of Recent Evidence

**Aya Mousa [1,*], Amreen Naqash [2] and Siew Lim [1]**

1 Monash Centre for Health Research and Implementation (MCHRI), School of Public Health and Preventive Medicine, Monash University, Melbourne VIC 3168, Australia; siew.lim1@monash.edu
2 Department of Pharmaceutical Sciences, University of Kashmir, Srinagar, Jammu and Kashmir 190006, India; anaqash.scholar@kashmiruniversity.net
* Correspondence: aya.mousa@monash.edu; Tel.: +61-385-722-854

Received: 10 January 2019; Accepted: 14 February 2019; Published: 20 February 2019



Poor dietary intakes or deficiencies in key macronutrients and micronutrients can therefore have a substantial impact on pregnancy outcomes and neonatal health

# FUNGSI MIKRONUTRIEN PADA PERIODE PERIKONSEPSI

**Table I Main functions of micronutrients involved in the periconceptional period**

| Micronutrient | Function | References |
|---|---|---|
| Folate | Involvement in the DNA replication (cell cycle); methylation cycle (amino acids cysteine and methionine cycle) | FAO/WHO Report (2004a), Scholl and Johnson (2000) |
| Vitamin $B_{12}$ | Conversion of homocysteine to methionine as cofactor of the methionine synthase | Ryan-Harshman and Aldoori (2008) |
| Vitamin $B_6$ | Metabolism of amino acids, lipids, one-carbon units and glycogen as co-enzyme; pathways of gluconeogenesis, heme and neurotransmitter biosynthesis | Mackey *et al*. (2006) |
| Vitamin A | Growth and differentiation of a number of cells and tissues | Ross (2006) |
| Antioxidants | Defence systems against free radical species | Fang *et al*. (2002) |
| Iron | Haematopoiesis; nucleic acid metabolism; carrier of oxygen to the tissues by red blood cell haemoglobin; transport medium for electrons within cells; integrated part of important enzyme systems | Cook *et al*. (1992); Beard (2003), FAO/WHO Report (2004c), Wood and Ronnenberg (2006) |
| Zinc | Structural, regulatory and catalytic functions as cofactor for numerous metalloenzymes | McCall *et al*. (2000), Hambidge (2000) |
| Copper | Neurotransmission, neuroropeptide maturation, oxidative phosphorylation, defence from free radical damage as cofactor for numerous cuproenzymes and copper-binding proteins | Turnlund (2006) |

# MIKRONUTRIEN DAN CACAT LAHIR

## FOLAT

Larut dalam air, banyak dalam sayuran hijau berdaun, ekstrak ragi dan buah jeruk

Berfungsi sebagai koenzim dalam transfer satu karbon selama siklus metilasi dan integral untuk sintesis DNA dan neurotransmiter

Defisiensi folat menghasilkan akumulasi homocysteine, yang dapat meningkatkan risiko dan kehamilan yang buruk termasuk preeklampsia dan anomali janin

Suplementasi dengan asam folat selama prakonsepsi dan kehamilan dini sangat penting dan dapat mencegah 40-80% dari cacat tabung saraf seperti spina bifida

# VITAMIN A

Vitamin yang larut dalam lemak berasal dari retinoid yang terbentuk sebelumnya atau provitamin karotenoid

Retinoid, seperti retinal dan asam retinoat, diperoleh dari sumber hewani termasuk telur, susu, hati, dan minyak hati ikan

Vitamin A tambahan diperlukan selama kehamilan untuk mendukung pertumbuhan dan pemeliharaan jaringan pada janin dan untuk menyediakan cadangan janin dan membantu metabolisme ibu

# Vitamin B

B-complex termasuk didalamnya vitamin B1 (thiamine), B2 (riboflavin), B3 (niacin), B6 (pyridoxine) and B12 (cyanocobalamin)

Vitamin ini bertindak sebagai koenzim di beberapa jalur metabolisme perantara untuk pembangkit energi dan pembentukan sel darah

Kekurangan riboflavin dan niasin telah dikorelasikan dengan preeklampsia, kelainan jantung bawaan, dan bayi BBLR

## ZINC

Kekurangan zinc pada awal kehamilan menyebabkan gangguan implantasi, aborsi dan malformasi janin, termasuk sumbing bibir dan langit-langit, malformasi otak dan mata, berbagai kelainan jantung, paru-paru dan urogenital sistem

## COPPER

Teratogenisitas terkait defisiensi tembaga seperti swollen hidbrains, penyakit lepuh, penggumpalan darah dan distensi pembuluh darah besar

# TAKE HOME MESSAGE

Cacat lahir adalah penyebab utama kematian bayi dan telah terjadi selama 25 tahun terakhir, menyebabkan 22% dari semua kematian bayi

Untuk memastikan awal yang sehat untuk bayi mereka, sebagian besar wanita sebaiknya menjalani beberapa bentuk skrining prenatal.

**Skrining trimester pertama akurat dan efisien dalam praktik klinis**

Kekurangan mikronutrien telah dikaitkan dengan risiko kehamilan yang sangat buruk, mulai dari infertilitas hingga cacat janin struktural dan penyakit jangka panjang


# TERIMAKASIH