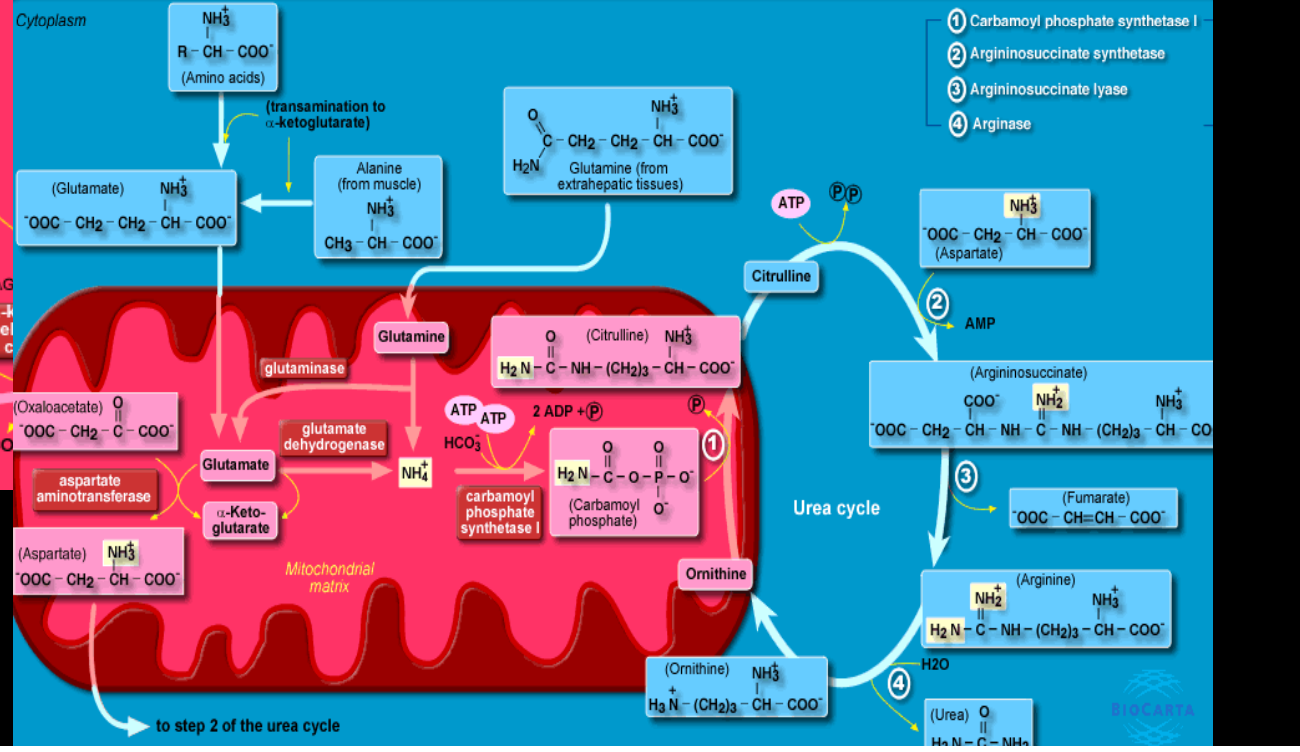
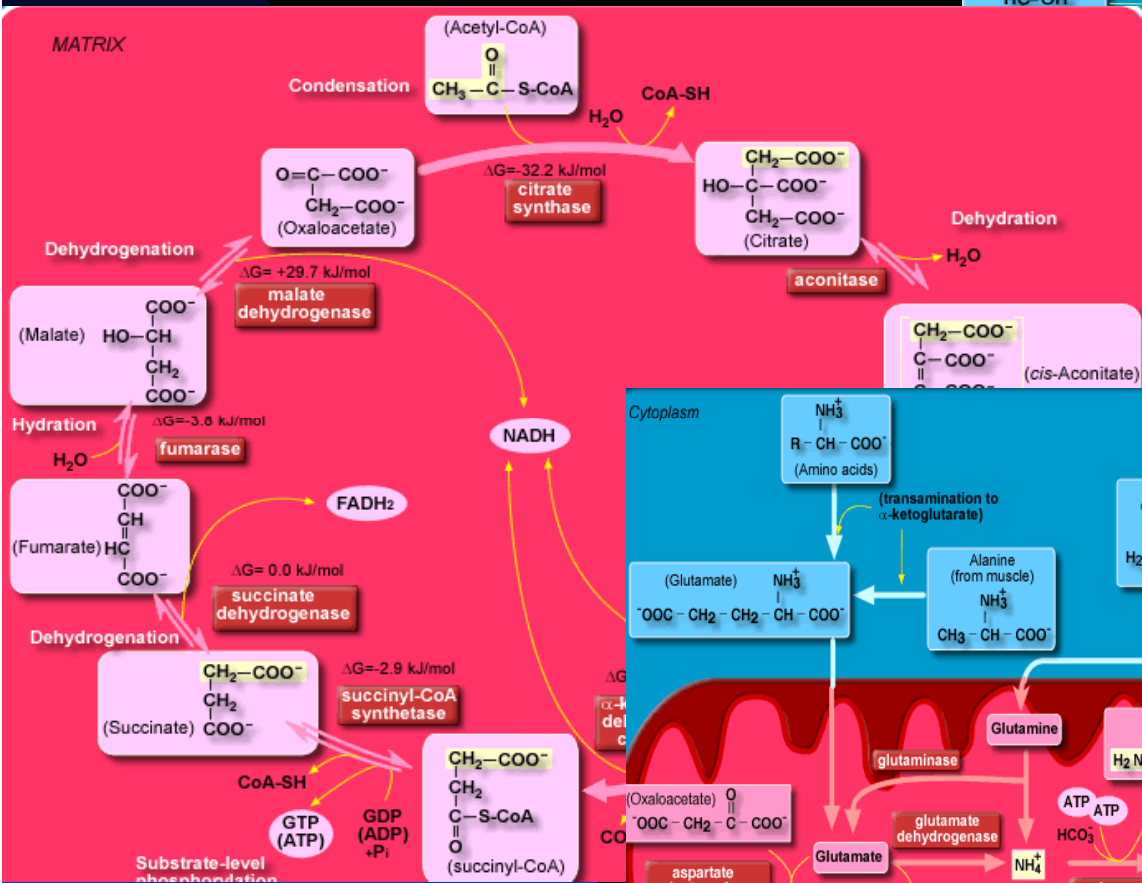
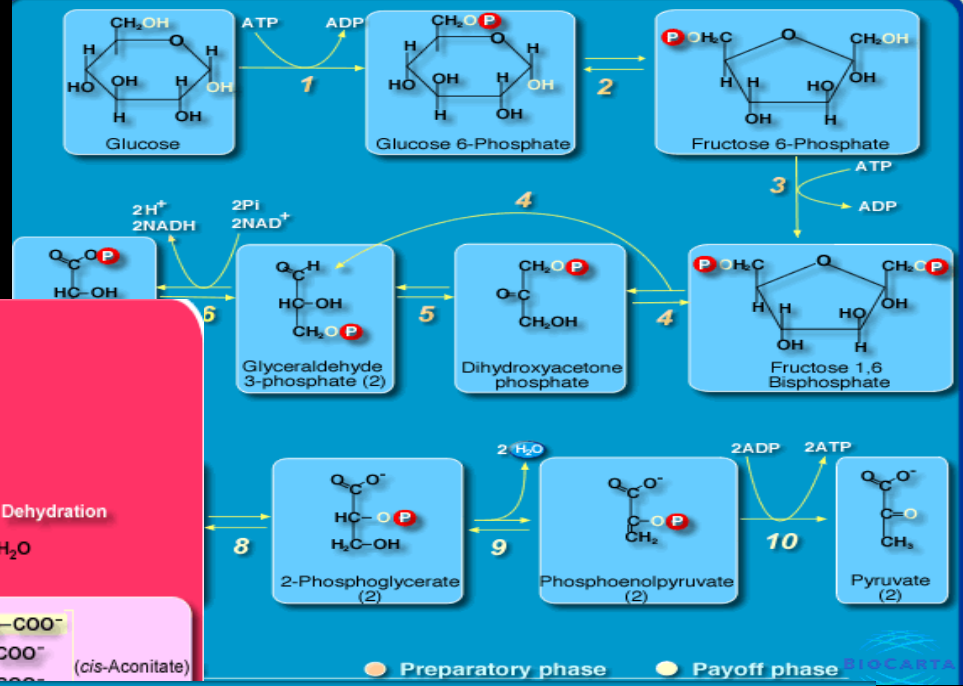


# Rare Disorders in Malaysia: Rare and Special

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Consultant Paediatrician and Clinical Geneticist  
Head, Genetic & Metabolism Unit,  
Department of Paediatrics,  
Faculty of Medicine,  
University of Malaya

# A Doctor's View of Diseases....



# Rare Disorders



“My Children, My Family,  
My Community !”



# Rare Disorders: Myths & Facts

- About 3-5% of babies are born with serious birth defects. Each year, there are about **20,000** Malaysian babies born with birth defects
- **Individually rare but collectively common**
- Some of these can be recognised at birth, while some are diagnosed later in life
- Some are due to genetic changes, while others are due to both genetic and environmental factors
- These conditions cause **medical and social** problems and may recur again in some families

# Rare Disorders:

Problems:

Low scientific knowledge (false perception about rare disorders - 'not important' enough)



Absence of cure and treatment that could improve quality of life



Low knowledge amongst health professionals & public



Negative social consequences

# What are Rare Disorders?

- Prevalence of less than 1 in 4000 people
- Examples:
  - ◆ *Osteogenesis imperfecta* (brittle bone disease),
  - ◆ *Duchenne muscular dystrophy* (DMD),
  - ◆ *Prader-Willi* syndrome,
  - ◆ *Fragile-X* syndrome,
  - ◆ Mucopolysaccharidosis
  - ◆ Chromosomal abnormalities
  - ◆ Inborn errors of metabolism
  - ◆ Syndromes and many others.

# Many challenges...

- Limited educational resources on rare disorders for patients and family members especially in different languages
- Lack of public support groups for patients with rare disorders
- Limited professionals trained in early intervention programmes and services
- Lack of awareness and understanding among medical professionals, organisations and community on the needs of individuals with rare disorders

# More challenges...

- Limited funds to support treatment or to purchase special orphan drugs
- Limited genetic testing available locally
  - too costly to send overseas
  - insufficient laboratories and technologists
- Limited local research data on rare disorders

# Day to day challenges...

- Specific diagnosis
  - ◆ Rare unique syndromes – parents feeling alone in facing challenges
  - ◆ Breaks for parents – respite care for caregivers
- Coping with loss of child, grief and bereavement
- Dealing with feelings of guilt, shame and blame
- No specific diagnosis:
  - ◆ dealing with uncertainties
  - ◆ management based on needs
  - ◆ family planning issues

# Day to day challenges...

- Health maintenance
  - ◆ Routine paediatric care e.g. vaccination
  - ◆ Monitoring growth and nutrition
  - ◆ Ensure development of skills
- Health surveillance
  - ◆ Hearing loss
  - ◆ Visual impairment
  - ◆ Difficulties in ambulating and getting around
  - ◆ Dental care
  - ◆ Personal hygiene and toilet

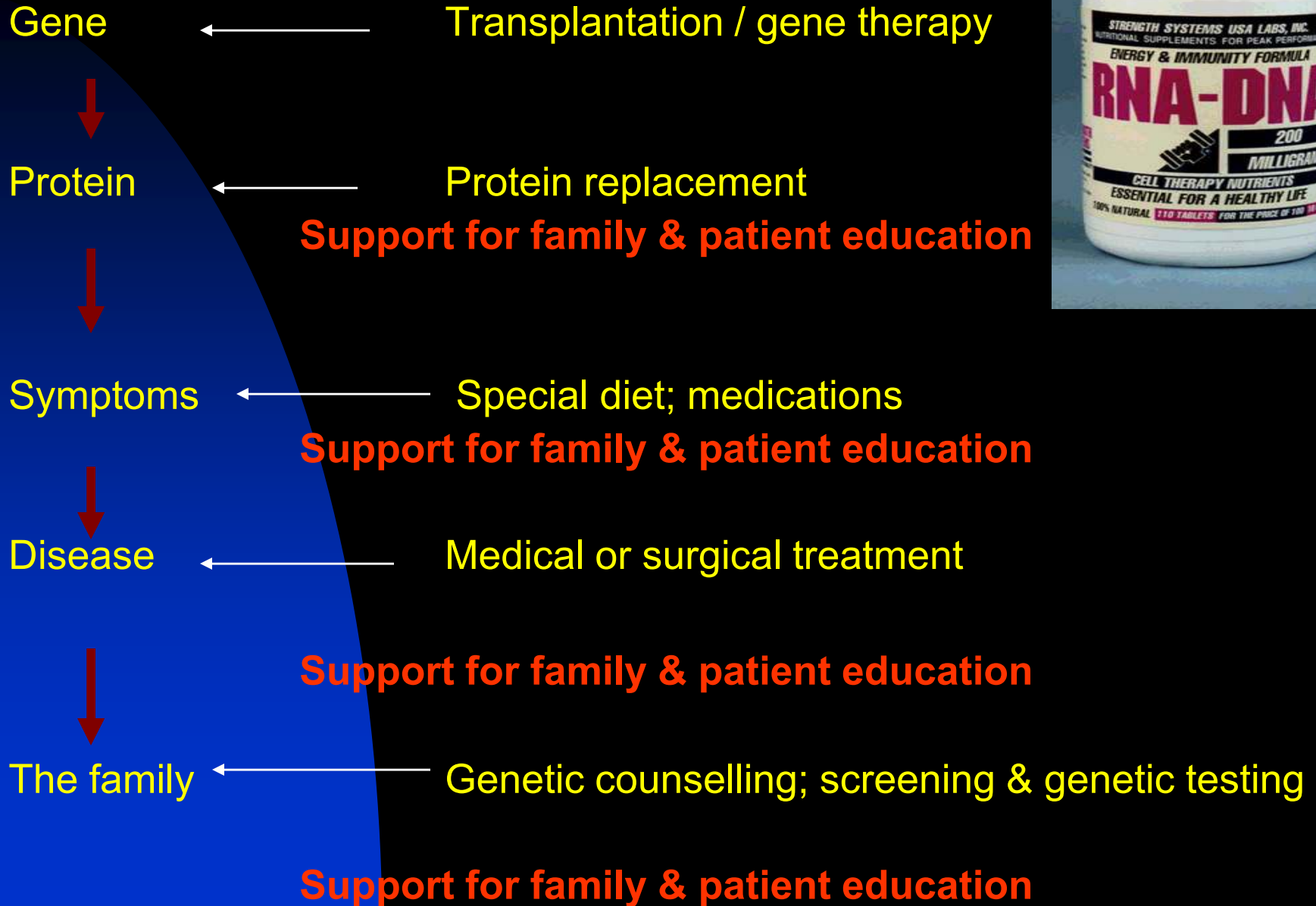
# More challenges...

- Sleep problems
- Behavioural challenges
  - ◆ Autism & Hyperkinetic disorder
  - ◆ Others: depression, aggression
  - ◆ Role of psychologist and psychiatrists
- Learning difficulties
  - ◆ IQ assessment: accurate?
  - ◆ Early intervention programme
  - ◆ Occupational therapy
  - ◆ Physiotherapy, speech therapy
  - ◆ Schooling?

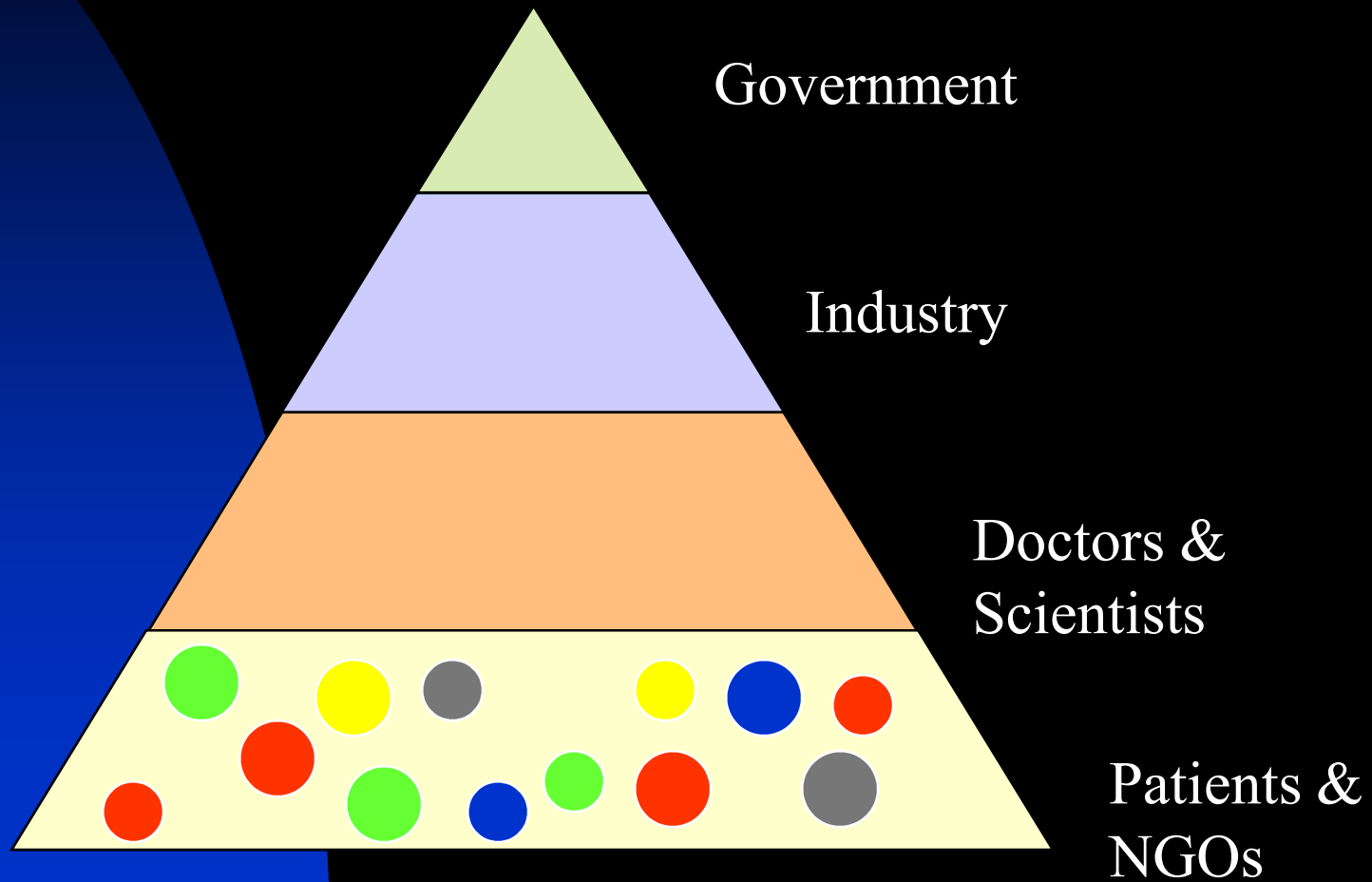
# Jabatan Kebajikan Malaysia

- [www.jkm.gov.my](http://www.jkm.gov.my)
- OKU registration
- Kemudahan pengangkutan awam
  - Tambang konsesi
  - MAS 50% penerbangan domestik
  - KTM 50% bagi semua kelas
  - LRT, KL Monorail 50%
  - Rapid KL – tiada bayaran
  - Transnasionaal 50%
- Pelepasan cukai sebanyak RM 5000 kepada pembayar cukai yang mempunyai anak OKU yang berusia dibawah 18 tahun
- Pelepasan cukai sehingga RM 5000 bagi membeli alat-alat khas untuk kegunaan sendiri, anak atau ibubapa OKU.
- Pengecualian Bayaran dokumen perjalanan

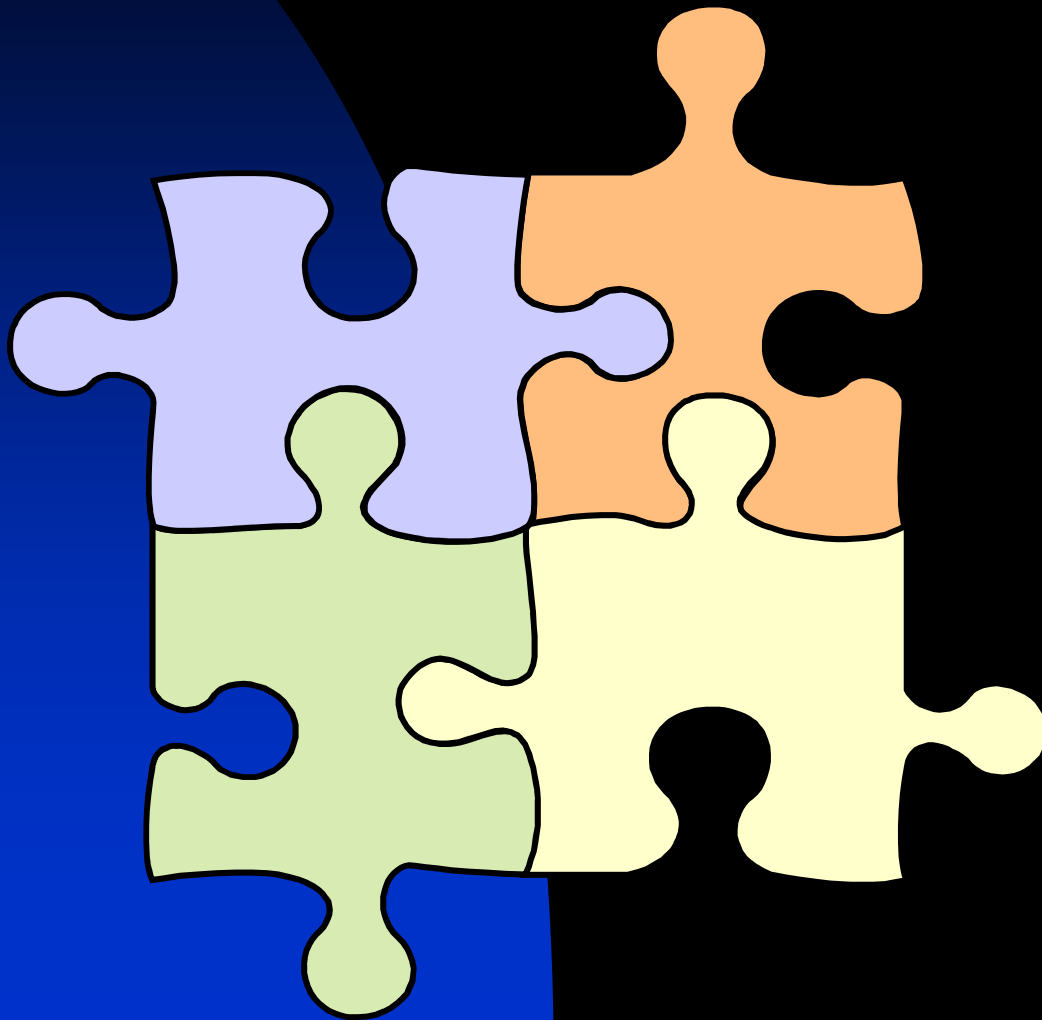
# Management of rare disorders



# Patients and their partners in health



# Rare Disorders: Putting the Jigsaw Puzzle Together



## Patient Groups:

Equal partners  
Share the burden  
Speak with one voice  
Work as a team  
Network together



**MALAYSIAN RARE DISORDERS SOCIETY**  
(Registration No. : 0064-07-WKL)



## Our hopes... Our dreams...

Create a network among individuals and families with rare disorders.

Create a network among MRDS members with organisations, agencies and professionals involved in treating, educating and conducting research on rare disorders.

Serve as a contact and resource centre on rare disorders.

Promote the awareness on rare disorders among the Malaysian community.

Support and assist individuals and families affected with rare disorders in terms of welfare, treatment, rehabilitation, education and social needs.

Collaborate with agencies and organisations that diagnose, research and treat rare disorders to increase the quality of life for the individuals and families affected.

Raise funds to support the activities and objectives of MRDS.

MRDS





# Thank you



# Our Team



**PRESIDENT : DATO' HATIJAH BINTI AYOB**  
**VICE-PRESIDENT : NOOR AZMI B ABU OSMAN**  
**SECRETARY : CHEW ANNA**  
**TREASURER : AHMAD AZAHAR BIN MOHD ZAIN**  
**COMMITTEE MEMBERS : MANIYARASU A/L PATCHIAPPAN**  
**ALLIDA MUHAMMAD SAID**  
**RAJESWARI A/P PONNUDURAI**  
**MRDS ADVISOR : PROFESSOR DR. THONG MEOW KEONG**  
**COORDINATOR : JULIANA LEE**