Genetics unit

Services

The unit provides diagnosis and treatment of various genetic and metabolic diseases in infants and children using various diagnostic tools including

- Karyotyping
- High resolution Karyotyping
- Survival motor neuron SMN gene : SMN1 And SMN2(Exon 7 and Exon 8)
- Familial Mediterranean Fever (MEFVgene :13 mutations by Real time PCR)
- Cystic fibrosis (CFTR : 8 common mutations,16 rare mutations by Real time PCR)
- Fragile X gene
- Azoospermia factor AZF gene testing for male infertility (Azospermia)
- HPLC (HBA, HBF, HBA2, HBS)
- Thrombophilia Factor II and V Leiden mutation analysis
- Enzyme assay, genetic testing, and enzyme replacement therapy for some lysosomal disorders as
  - -Mucopolysaccharidosis type I (**Aldurazyme)**
  - - Mucopolysaccharidosis type II (**Hunterase)**
  - - Mucopolysaccharidosis type IV (**Vimizim)**
  - - Mucopolysaccharidosis type VI (**Naglazyme**)
- Enzyme assay, genetic testing, and enzyme replacement therapy for Glycogen storage disease Pompe (**Myozyme**)
- Nutritional treatment of some disorders of inborn errors of metabolism:
  - -Phenylketonuria (**PKU** 1,2,3 and **Loprofin mix**)
  - -Glutaric acidemia (**XLYS, low Try Maxmaid**)
  - -Methyl Malonic acidemia (**MMA /PA infant anamix)**
  - - Maple urine syrup disease (**MSUD anamix**)
  - -Protein free-High Caloric **(Ducal):**
    - *Hyperammonemia
    - *Urea cycle defects
  - low in long chain triglycerides (LCT) and high in medium chain triglycerides (85%)) **(Monogen)** Indicated in:

✓ long chain fatty Acid oxidation
✓ Chylothorax.
✓ Cystic fibrosis.
✓ Hyperlipoprotienemia type 1.
✓ Intestinal lymphangiectasia.

- ✓ Intractable malabsorption with steatorrhea.
- ✓ Post-operative feeding in short bowel syndrome.
- ✓ Other lipid and lymphatic disorders where a low fat, and/or low LCT, high MCT diet is indicated.

- **Ketocal** 3:1(fat : carbohydrates ₊ protein) indicated in:
    1. Pyruvate dehydrogenase deficiency
    2. Glucose transporter type -1 defeciency
    3. Intractable Epilepsy

- Treatment of acute hyperammonaemia and acute decompensations of UCD

  *Carglumic acid (**Carbaglu**®)

Staff members:

Prof. Mohamed Abdelrahman Shokeir

Prof. Ali Ali Ibrahim Shaltout

Prof. Mohamed Mohamed Saleh Elhaggar (Current head of the unit)

Prof. Soheir Yehia Abdelrazik Mohammed

Assistant Prof. Dina Mohammed Abdelhady

Assistant Prof. Yahia Mohammed Mokhtar Abdelrahman

Assistant Prof. Abdelrahman Eid Mahmoud

Lecturer Ranim Mohammed Hassanin
Assistant Lecturer Heba Eltaher
Assistant Lecturer Zahraa abdelmonem

Prizes:

Prof. Soheir Yehia (Award of Prof. Ekram Abdelsalam in medical genetics 2019)

Dr. Ranim Hassanin (Award of Prof. Yehia Elgamal in Pediatrics presented from academy of scientific research at 2020)