

MULTIDISCIPLINARY MANAGEMENT AS A RESOURCE FOR METABOLIC DISEASES:



COOPERATION BETWEEN CLINICIANS AND PHARMACISTS IN AN ITALIAN CENTRE FOR RARE DISEASES

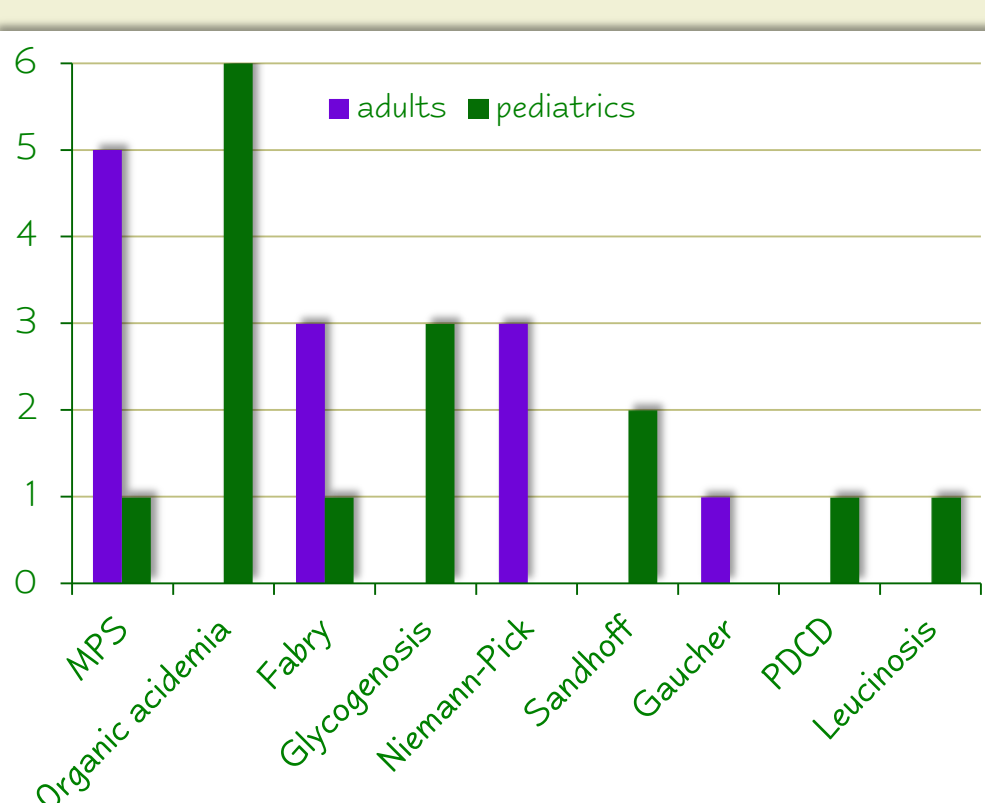
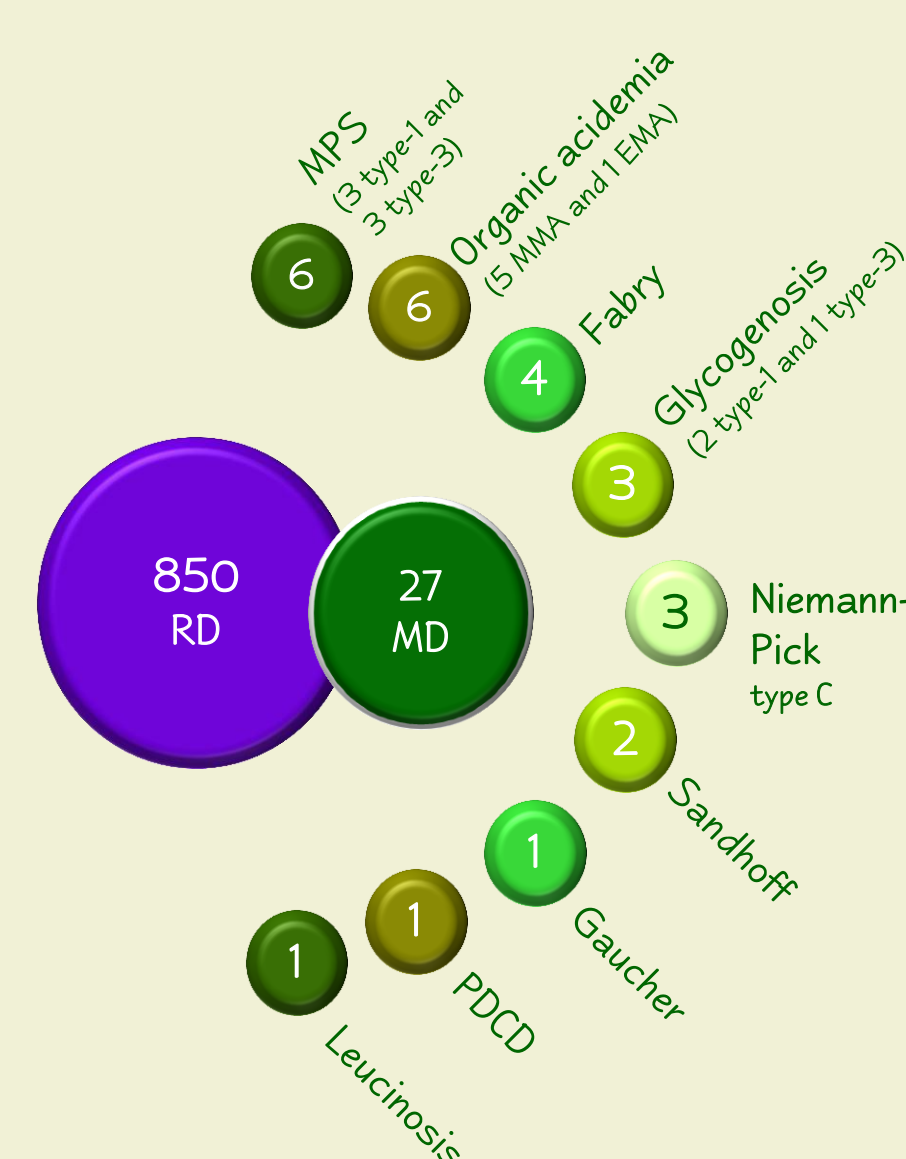
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Picture 1: Frequency of Metabolic Disease (MD) per type of patient (adult or pediatric)



BACKGROUND

Rare diseases (RD) are almost 8.000 (80% genetically based) and 50% of them affects pediatric age.

Their complexity is extremely relevant upon metabolic diseases (MD): heterogeneity (age of occurrence, etiopathogenesis and symptomatology), low occurrence, late diagnosis, lack of resolutive therapies and multisystemic involvement.

The Decree 279/2001 of Italian Minister of Health institutes the "Rare Disease National Web" and assigns the Regions the task to find hospitals and organizations to supply health services. Since 2002 our hospital "G. Salesi" is the reference centre for Marche Region.

PURPOSE

The cooperation between doctors, pharmacists and health workers, which was spontaneously born of individual needs and emergencies, needed to guarantee optimized treatments, respond to therapeutic/welfare necessities, and treat patients' extremely complex urgencies.

MATERIALS and METHODS

We verified through the analysis of medical records: patients with MD who enter our centre (diagnosis, therapy and follow-up) and prescriptions (basic therapy/emergency support). Hospital and territorial doctors and pharmacists met in order to layout shared protocols.

RESULTS

Our Centre treats 850 RD-patients. MD-patients are 27 (13 males and 14 females) and 59% is pediatric (10 patients <10 years old and 6 ≥ 10 years old) (Picture 1). We care 9 types of MD: Fabry (4), Sandhoff (2), Gaucher (1), mucopolysaccharidosis (MPS) (6), organic acidemia (i.e. methylmalonic-MMA and ethylmalonic-EMA) (6), glycogenosis (3), leucinosi (1), Niemann-Pick type-C (3) and pyruvate dehydrogenase complex deficiency (PDCD) (1) (Picture 1). Seven of our patients received MD-diagnosis from other hospitals. 30% of our patients needs a specific diet regimen and 18% of them needs galenic preparations (Picture 2 and Table 1). Five patients follow the enzymatic therapy in day-hospital: 2 (Iaronidasi) 2 (agalsidase alpha), 1 (idursulfasi). 26% of diagnosis comes from other centers.

We wrote up a protocol to manage with all the dietetic and therapeutic emergencies and sent it to all operators, especially for emergencies for metabolic acidosis, hyperammonemia, and methylmalonic aciduria.

CONCLUSION

The "doctor-pharmacist" team was born to improve MD-patients' quality of life. The pharmacist's role consists of: respond to lack of resources/cures (i.e. galenic preparations suitable for pediatric age), help doctors in therapeutic decisions, support patients and families in sanitary assistance.

Picture 2: MD treatment (diet or pharmacological)

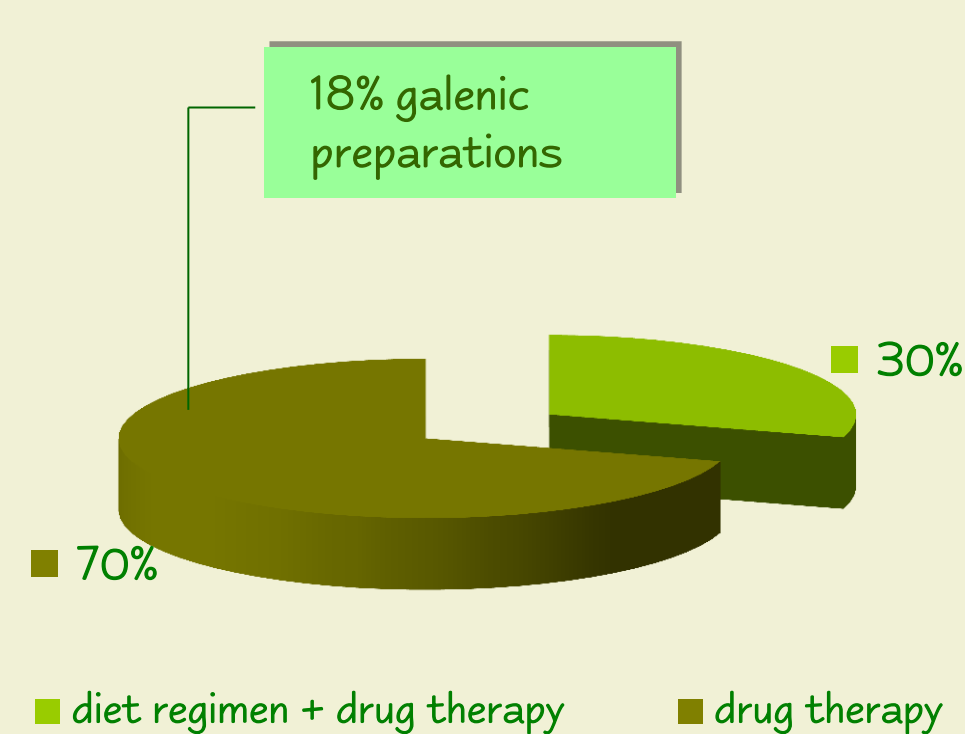


Table 1: Galenic preparations requests to pharmacists

Galenic preparations	Metabolic Disease
potassium citrate syrup 1 mEq/ml; sodium dichloroacetate powder 50 mg	PDCD
riboflavin caps 50 mg	EMA
levofoinic acid caps 0,5 mg; modified Shohl's solution 1 mEq/ml	MMA
miglustat caps personalized dosage (from 11 mg to 70 mg); potassium citrate syrup 1 mEq/ml; potassium bromide syrup 50 mg/ml	Sandhoff