



A case of Mitchell Riley Syndrome: when the team's work is winning!

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Mitchell Riley syndrome

- Is a **rare genetic** disorder
- It is presented with a triad of severe neonatal **diabetes** (pancreatic hypoplasia), **gallbladder agenesis/hypoplasia**, and **intestinal atresia**
- The **prognosis is poor**

Prognosis in the previously reported cases compared to the current patient

Prognosis		
Mitchell et al. (1)	1	Death at 5 months of age.
	2	Death at 6 months of age.
	3	Alive at 9 years, intermittently off insulin.
Chappell et al. (9)	4	Alive at 6 years, on an insulin pump, diabetes well controlled, thriving well, developmentally appropriate to age.
Martinovici et al. (3)	5	Death at 2 months of age.
Smith et al. (10)	6	Death at 2.5 months of age.
Spiegel et al. (4)	7	Alive at 1 year and 9 months, on insulin 0.7 U/kg/day, diabetes well controlled, HgA1c 7.1% persistent diarrhea on home TPN, moderate motor delay.
Concepcion et al. (11)	8	Death at 5 months of age.
Chandra et al. (5)	9	Alive at 6 years, on inulin 0.6 U/kg/day.
Sansubury et al. (6)	10	Alive at 9 years, on insulin 0.35 U/kg/day, HgA1c 8.96%
	11	Alive at 9 years, on insulin 0.7 U/kg/day, diabetes well controlled HgA1c 7.2%
Zegre Amorim et al. (8)	12	Alive at 7 months, on an insulin pump with malabsorption. On home TPN.
Khan et al. (7)	13	Not reported.
Skopkova et al. (2)	14	Alive at 13 years, on insulin 1.0 U/kg/day, diabetes well controlled, HgA1c 7.3%, developmentally well.
	15	Alive at 8 years, on insulin 0.95 U/kg/day, diabetes well controlled, HgA1c 6.9%, developmentally well.
This report	16	Death at 5 months of age.

*Mitchell-Riley syndrome report of novel mutation and review of the literature
Nourah Alruqaie et al, Journal of Biochemical and Clinical Genetics 2018; 1(2):87-92*

Case presentation

- a male Pakistani infant, born of first-cousin parents, low weight for gestational age, no dysmorphic features.
- Clinical features: severe neonatal diabetes (pancreatic hypoplasia), duodenal atresia, malabsorption, chronic diarrhea.
- **Hospitalized for seven months** in a Neonatal Intensive Care Unit

Patient's needs

- Physical: malabsorption, chronic diarrhea, electrolyte imbalance (required **NPT**)
- **central venous catheter**
- diabetes (**insulin pump**)
- Psychological: need of a family environment

Family's needs

- Educational: caregiver training
- Psychological/Comunicative: cultural-linguistic barrier

Social, financial and health insurance

The answer to complexity....



...was a paediatric palliative care service that isn't available in our local reality.

Thus We transferred the patient to a Paediatric Hospice for

- ✓ support
- ✓ caregiver training

During this period the child developed two central venous catheter sepsis, one of which required hospitalization in a Paediatric Intensive Care Unit.

Clinical stability

- ✓ After ten months of hospitalization, the child reached a period of clinical stability.
- ✓ The mother expressed the need to get closer to the family.
- ✓ The collaboration was born between the colleagues of the Paediatric Hospice and those of the child's hometown.



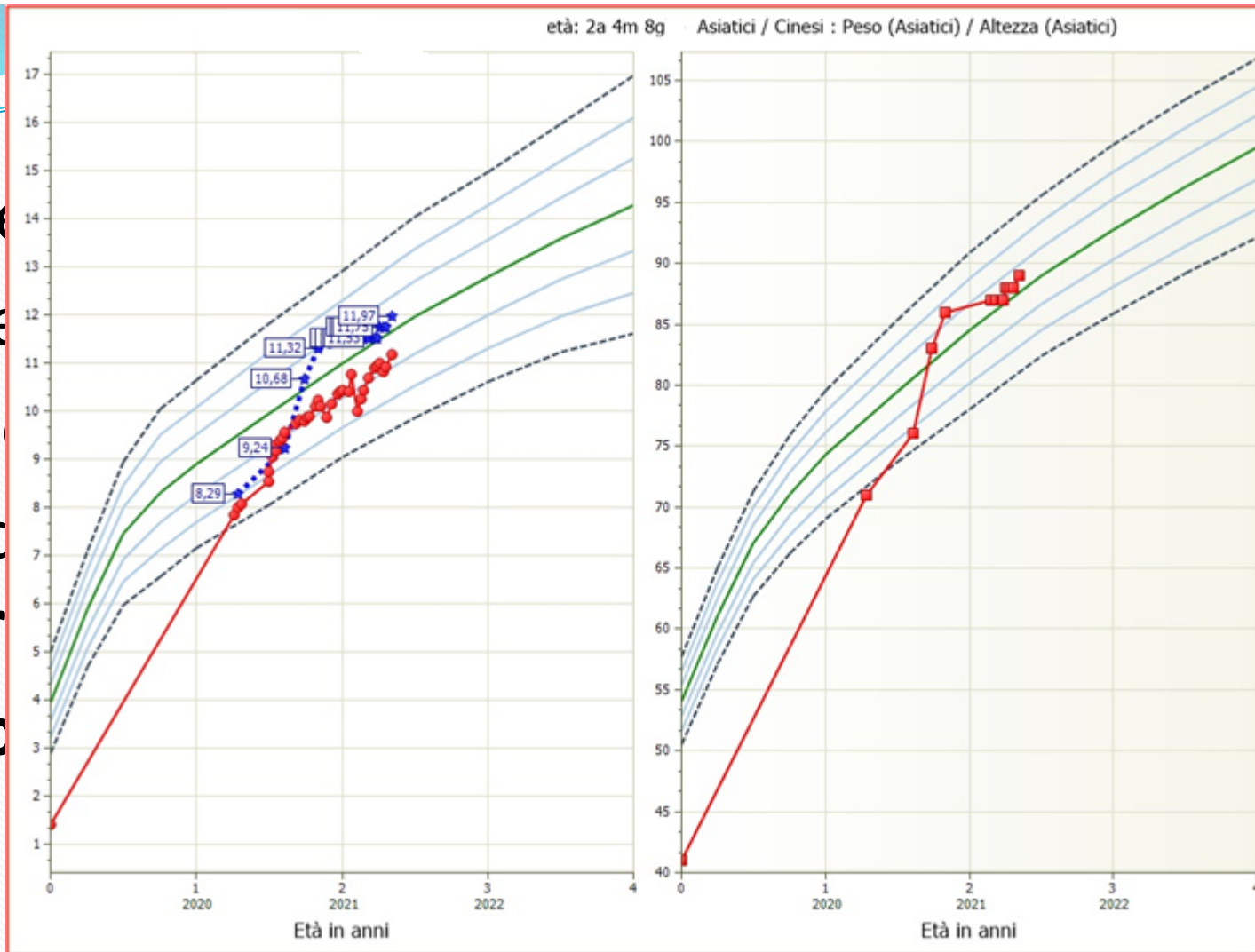
The **team** is composed of:

- Hospital doctors (among them an expert in CPP)
- Hospital nurses in charge of local 'protected discharge', who facilitate a smooth transition from hospital to home care.
- Family Pediatrician
- Home nurses and their territorial case manager
- Cultural mediator
- Local volunteering association



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Conclusion: teamwork is winning!

Despite clinical care complexity, socio-cultural and family difficulties, integrated work can guarantee a prolonged survival, with a good quality of life for children with Mitchell Riley syndrome.





 Thank
you

