

# Familial rectal pain: a familial autonomic disorder as a cause of paroxysmal attacks in the newborn baby

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**ABSTRACT** – A 2-day-old baby exhibited impressive paroxysmal attacks consisting of bradycardia, bronchospasm and vasomotor fits (Harlequin type) related to a rare, dominantly inherited form of dysautonomia called “familial rectal pain”. These events were recurrently triggered by emotion, diaper changes or wiping of the perineal areas or eating. Sometimes they occurred spontaneously. Carbamazepine had an excellent effect on the fainting. At four years of age, the child had normal psychomotor development with only minimal symptoms, and very rare paroxysmal attacks. The father of the child has minimal symptoms of this entity, with essentially ocular manifestations. Familial rectal pain is a very rare entity that must always be considered as a possible aetiology of any life-threatening event in an infant because of the availability of a very effective treatment. The existence of minimal forms of familial rectal pain is possible, and it is likely that this entity is underdiagnosed.

*[Published with video sequences].*

**Key words:** Newborn, Dysautonomia, Bronchospasm, Differential diagnosis of epilepsy, Carbamazepine

Syncope is an unusual symptom in the term newborn that requires extensive investigation in order to identify cardiac or gastro-intestinal disorders, or epileptic seizures. We were puzzled by a term newborn who presented, on the second day of life, impressive syncopes with bronchospasm and bradycardia. The flushing observed had peculiar characteristics marked by paroxysmal vasomotor changes to half of the body (Harlequin type), which persisted after fainting. Between these acute events, clinical and neurological examinations were normal. Full investigations led to the diagnosis of a familial dysautonomia called “family rectal pain”. We review the differen-

tial diagnoses that should be discussed in the presence of such symptoms, and the therapeutic consequences of the diagnosis.

## Case report

This full term, male infant, the first child born to healthy parents with no family antecedents, suffered from paroxysmal events at two days of age. These impressive events started with tonic posturing followed by bronchospasm apnoea and severe bradycardia, and which were recorded on the video sequence (see video sequence 1). These attacks occurred either spontaneously or following



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bowel movements. A typical episode consisted of flushing, initially of the whole body and then one half of the body, with loss of consciousness following syncope and bronchospasm that lasted 10-30 seconds. Some flushes lasted 10 to 30 minutes. On each occasion, flushing displayed a different topography. The demarcation between erythematous and adjacent skin was sharp (see video sequences 1 and 3). Between attacks, neurological development was normal (see video sequence 5).

Aetiological investigations included esophageal pH probe studies and gastrointestinal barium contrast X-ray; neurotransmitter levels (in cerebrospinal fluid and urine) were normal and toxicology screen negative. EEG activity showed no change during the paroxysmal event. Nasal percussion failed to trigger syncope, and clonazepam had no effect. Intradermal reaction was positive. MIBG and technetium scintigraphies were normal. The diagnosis of "familial rectal pain" (FRP), a rare familial dysautonomia, was made based on the typical aspects of the paroxysmal events.

Carbamazepine was extremely effective, reducing severe attacks to once a month or once a trimester during the second year of life. On follow-up, the patient had had severe rectal pain with bronchospasm (obstructive apnoea) essentially during the first 18 months of his life. Falls on the buttocks delayed independent walking (22 months). He developed ocular manifestations such as excessive weeping with or without rectal pain attacks. Submaxillary pain occurred infrequently after 18 months of age, and was triggered by the sight of food. These symptoms persist today.

At five years of age, his neuropsychological development was normal, and severe attacks were very rare. Some minimal events had occurred, such as cheek flushing or conjunctival suffusion, as in his father's case, especially when he saw certain types of food. These attacks were partially controlled by amitriptyline. Then, he developed arterial hypertension treated with dual antihypertensive therapy.

Retrospectively, we learned that the child's father had presented with some conjunctival suffusion and flushing of the eyelid and the skin in the temporal area, lasting for a few minutes. Later, when seeing certain food, he also experienced bursts of pain in the parotid region affecting both sides which was associated with transient erythematous skin and salivation. He had never presented with an attack as severe as his son's.

## Discussion

"Familial rectal pain" was first described in 1959 by Hayden and Grosman as an autonomic disorder which, in its severe form, generates its first symptoms in the neonatal period. Clinical symptoms are mainly rectal pain, ocular and submaxillary flushing (Hayden *et al.* 1959, Emslie *et*

*al.* 1996, Dugan *et al.* 1972, Mann *et al.* 1972). The aetiology and pathophysiology of the condition are unknown. The symptomatology suggests an abnormality of the autonomic nervous system. The neurological course is favourable.

The expression of the condition may vary. Some affected individuals have prominent ocular and submaxillary symptoms, while others have mainly rectal symptoms. This familial dysautonomia is an autosomal dominant condition, with varying degrees of penetrance (Hayden *et al.* 1959, Dugan *et al.* 1972). Ocular and submaxillary symptoms seem to have a higher degree of penetrance than the rectal symptoms (Hayden *et al.* 1959).

Features of rectal pain attacks consist of tonic contraction of the whole body, with bronchospasm and bradycardia followed by tachycardia with colour changes (Mann and Cree, 1972). These episodes may be precipitated by defecation, insertion of a rectal thermometer or a fall on the buttocks. According to the carer's description, an episode begins with the sudden onset of an intense sensation of burning in the perineal region that spreads rapidly. Pain is accompanied by flushing of the lower half of the body, variably affecting either one side or the other, or both. Pain appears suddenly and lasts from 10 seconds to one minute. Affected persons report that this pain is the worst they have ever experienced. It almost invariably accompanies or immediately follows bowel movement. Immediately after the pain has subsided, skin on the buttock and genitals reddens. Affected persons have a normal life and usually suffer from no other disability except for frequent jaw ache. They invariably develop chronic constipation. During childhood and over the years, sufferers gradually learn to control bowel movements but defecation, digital examination of the rectum, coitus and parturition commonly trigger attacks in adults. The frequency of attacks tends to decrease on approaching adolescence.

Ocular and submaxillary symptoms are often associated with rectal symptoms. Ocular pain with excessive watering caused by cold wind or other irritant factors, and jaw pain associated with local flushing triggered by the sight of food may occur later in the course of the disease and persist for life. Episodes of ocular and submaxillary pain are sometimes experienced from infancy. Pain is then described as needle-like, but lasts 10 seconds and is much less severe than the rectal pain. Blurred vision with flushing of the lid and periorbital skin may be associated. Submaxillary pain is described as sudden, and radiating over one side, being usually induced by the sight of food. It is immediately followed by a few seconds of profuse, watery salivation.

A neonatal case was reported in which the first attack occurred following a smack to the buttocks given by the midwife to stimulate crying and breathing: a red flush spread over the baby's buttock and down the back of the leg to the sole of the foot (Mann and Cree, 1972). Neonatal episodes with rectal symptoms and bronchospasms are

the hallmark of severe FRP. The most effective treatment is carbamazepine. In our patient, this drug reduced the frequency of episodes, but the attacks also seem to be becoming less frequent and less severe, as the child grows older. Amitriptylin (Dr Stephenson's personal communication) and gabapentin (Dr Aicardi's personal communication) are also effective. Differential diagnoses were:

- Sandifer syndrome due to gastrointestinal reflux. This diagnosis was excluded by the usual oesophageal pH probe studies and gastrointestinal investigations. Epilepsy could be excluded because the ictal EEG was normal, and antiepileptic drugs (valproate, benzodiazepines) were not effective;

- Hyperekplexia (stiff man syndrome) was considered, but examination was normal and nasal percussion failed to trigger fits. This manoeuvre suggested by Vigeveno *et al.*, (1989) consists of nasal percussion, which provokes a sudden hypertonia with or without apnoea plus bronchospasm. This is considered as pathognomonic in this condition. Clonazepam, an effective therapy in hyperekplexia, had no effect in this case;

- Riley-Day's familial dysautonomia was excluded by the absence of hypotonia, the presence of deep tendon reflexes, and the normality of the pupil response, while the intradermal reaction (histamine test equivalent) was positive, excluding a sensitive nerve disorder;

- Apudomas such as pheochromocytoma, mastocytosis or carcinoid tumour could be ruled out based on normal hormonal and neurotransmitter levels. MIBG and technetium scintigraphies were normal;

- "Familial rectal pain: a type of reflex epilepsy?" was hypothesised by Schubert *et al.* (1992). Two members of a family suffering from FRP had ictal EEG recordings showing only flattening and slowing of the trace. This pattern was not characteristic of an epileptic event and could be seen during an anoxic episode reflecting a cerebral hemodynamic phenomenon. Thus, antiepileptic therapy was ineffective, apart from carbamazepine and gabapentin which have been found to be effective in FRP. The epileptic nature of the manifestations in Schubert's case remains controversial;

- Trigeminal neuropathy shares common features with FRP: pain and flushing of one side of the face occasionally accompanied by conjunctival suffusion. It has the appearance of a local form of FRP, and symptoms are also relieved with carbamazepine therapy.

## Conclusion

Familial rectal pain syndrome apparently represents a disturbance of the autonomic nervous system, with somatic pain and autosomal dominant transmission. It is rare but must be considered as a possible aetiology of any newborn and infant paroxysmal event. It is important to recognize this syndrome because an effective treatment

## Legend for video sequences

*Video sequence 1* (Gauthier, 3-weeks-old): the baby presents with faintness while breastfeeding, with initially a cry, and afterwards exhibiting a generalized hypertonia, diffuse redness and loss of consciousness. The baby stops breathing (bronchospasm). The child begins to regain consciousness as the vasomotor phenomenon appears. At the end of the sequence, the neck is colourless, the face and the thorax are red. During the paroxysmal attack no changes are seen on the EEG.

*Video sequence 2* (Gauthier, 3 months old): at the beginning of the episode, the baby suddenly cries and rapidly becomes unconscious. He stops breathing (bronchospasm), has general flushing of the face, becomes cyanotic and then extremely pale.

*Video sequence 3* (Gauthier, 5 months old): the child exhibits vasomotor changes seen after recovery from the bronchospasm which persist for twenty or more minutes after the attack. Notice the precise demarcation between one half of the face and the other.

*Video sequence 4* (Gauthier, 5 months old): the episode begins with severe pain. Notice the beginning of the attack showing a suffering baby who becomes unconscious. Notice also the vasomotor changes to the face, especially the redness of the upper left face. The video shows that the baby is in severe pain probably with severe paroxysmal headaches occurring with the vasomotor changes. No EEG modification is seen.

*Video sequence 5* (Gauthier, 5 months old): good neurodevelopment. The baby performs well socially and has good axial tonus.

exists that very quickly relieves the pain experienced by the infant. Minimal forms of FRP exist. □

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