More than meets the Smile: Facial Muscle Expression in Children with Ochoa Syndrome

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SUMMARY

The Ochoa syndrome is the association of a non-neurogenic neurogenic bladder with abnormal facial muscle expression. Patients are at risk for renal failure due to obstructive uropathy. We report a family of three siblings, with an emphasis on the abnormalities in facial expression. Careful examination shows an unusual co-contraction of the orbicularis oculi and orbicularis oris muscles only when full facial expressions are exhibited, across a range of emotional or voluntary situations. This suggests a peripheral disorder in facial muscle control. Two thirds of patients have anal sphincter abnormalities. Aberrant organisation of the facial motor and urinary-anal sphincter nuclei may explain these symptoms.

KEY WORDS:
Abnormal facial expressions, Neurogenic bladder, Ochoa syndrome

BACKGROUND

The Ochoa syndrome describes the combination of a non-neurogenic neurogenic bladder in association with abnormal facial muscle expression [1]. More than 100 cases have been reported to date, mainly describing the urological and renal characteristics of the syndrome. Little has been written about the abnormalities in facial expression, which greatly aid the nephrologist or urologist in making the diagnosis. In this article we aim to explore the characteristics of facial muscle expression in these patients.

CASE REPORT

We report a family in which three siblings have Ochoa syndrome: a 17 year old boy and two younger sisters aged 11 and 8 years old respectively. The parents are distantly related. All three share the classic features of Ochoa syndrome - peculiar facial expressions and dysfunctional voiding. The older girl also has anal incontinence when attempting to micturate, needing to manually occlude the anus by squeezing her buttocks together whilst micturating.

The youngest is the index case and has been more thoroughly evaluated, as she presented at age 3 years with urinary tract infection and renal impairment. She has never been able to void fully on her own. She feels a sensation to void but is only able to pass small amounts of urine. Urinary stream is poor, and there is a constant dribbling of urine. Residual urine volume is usually large. She has been compliant to regular intermittent catheterization and prophylactic antibiotics.

Neurological examination of lower limbs is completely normal; there is no spinal dysraphism. Ultrasonography showed a distended bladder with bilateral moderate hydronephrosis. Micturating cystourethrogram revealed a trabeculated bladder with diverticulum and grade III reflux into the left ureter but no reflux into the right side (Figure 1). Urodynamic studies showed high opening pressures with a high residual volume of 220ml. The MRI of the spinal cord was normal. The diagnosis of Ochoa Syndrome was suspected when she was noted to have repeated episodes of peculiar facial grimacing during the clinic review. On further probing, it came to light that this was her typical facial appearance when she attempted to smile.

Figure 2 (i, ii) shows the facial expressions of the 11 and 8 year-old sisters, across a range of voluntary tasks. All three children share the same features. The abnormalities are seen

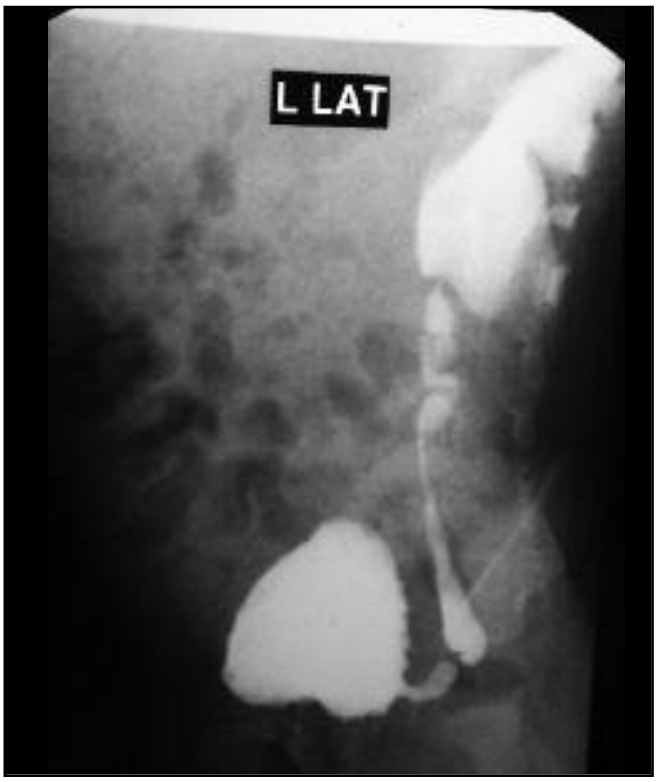

Fig. 1: Micturating cystourethrogram showing a trabeculated bladder with diverticulum and grade III reflux into the left ureter.

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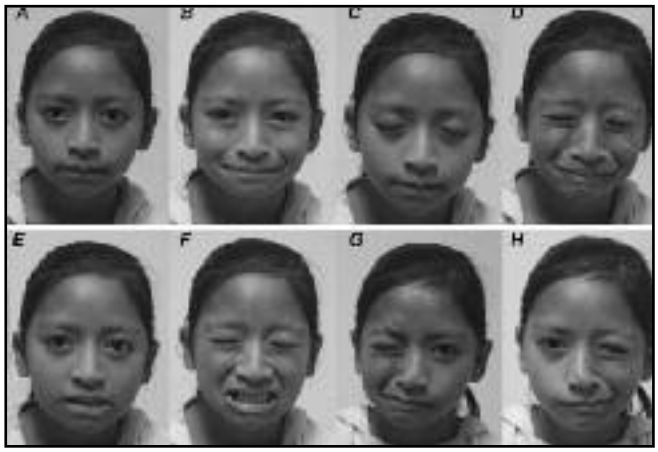

Fig. 2: (i) Elder sister. Plate A-D: Smiling A, Face at rest; B, Half smile – normal expression; C, Half smile with eyes closed; D, Peculiar facies: a contorted grimace emerges with full smiling.
Plate E-H: Other Facial Expressions: E, Showing teeth: normal expression; F, Peculiar facies of grimace emerges when asked to show teeth fully; G,H. Scrunching the face on the right and left sides show a partial grimace on either side.

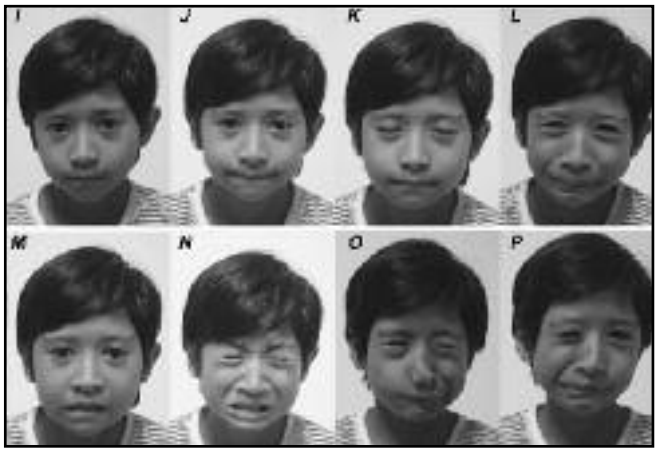

Fig. 2: (ii) Younger sister. Plate I-L: Smiling: I, Face at rest; J, Half smile – normal expression; K, Half smile with eyes closed; L, Peculiar facies: a contorted grimace emerges with full smiling.
Plate O-P: Other Facial Expressions: M, Showing teeth: normal expression; N, Peculiar facies of grimace emerges when asked to show teeth fully; O,P. Scrunching the face on the right and left sides show a partial grimace on either side.

with voluntary, emotional and reflex facial movements. Their four normal siblings and parents do not exhibit this abnormality.

DISCUSSION

A loss of function mutation in the *Heparanase 2 (HPSE2)* gene on chromosome 10q24 is the cause of Ochoa Syndrome, encoding a 592 amino acid protein with a yet undefined biological function [2]. All patients have a neurogenic bladder in the absence of a demonstrable urinary outlet obstruction of spinal cord pathology, thus resulting in a "non-neurogenic neurogenic bladder". The peculiar facial features are seen in all patients, and helps differentiate this disorder from other causes of a non-neurogenic neurogenic bladder, such as Hinman syndrome [3]. MRI studies have revealed normal brain and spinal cord structures in all patients thus far [1].

A careful examination of our patients (Figure 1) show normal facial expression across a wide range of range of emotions and voluntary control. However, when the expressions involve a large contraction of the orbicularis oris muscle, there will be concomitant co-contraction of the orbicularis oculi muscle as well, resulting in a peculiar facies. This peculiar facial grimace occurs with both voluntary and emotional facial expression. Control of facial musculature is represented by five cortical regions, but situated in two different brain regions: the primary motor cortex and an adjacent premotor cortex are lateral structures in the frontal lobe, and three facial motor cortices at the cingulate gyrus lie in the medial frontal lobe.

These areas exert control over the facial musculature differently in terms of location (upper/lower face) and under different circumstances (emotional vs. voluntary facial expressions) in a much more complex manner than was previously understood [4]. Patients with isolated lesions in either one of these regions produce clinical syndromes of dissociative emotional and voluntary facial function - being unable to voluntarily smile but paradoxically having a spontaneous, full smile in response to humor, or vice versa [5]. In our patients, the unusual facial expression is seen with both volitional and emotional circumstances, thus suggesting a more peripheral disorder in facial muscle expression.

The facial cortices control distinct patterns of facial muscle expression via the facial motor nucleus at the pons. This nucleus has four subnuclei which control different facial muscle groups separately: namely the occipitofrontalis (scalp and upper face), the auricular muscles and the muscles around the mouth [4]. Thus, the orbicularis oculi and orbicularis oris muscles are innervated by different subnuclei. In our patients (Figure 1), milder facial expressions appear normal, but full expressions appear to recruit both the orbicularis oculi and orbicularis oris, regardless of the initially intended expression. Possible explanations for this could be a disordered innervation of the facial motor nuclei by the facial cortex, or a disordered organisation of the facial motor nucleus, in which a subnuclei innervates both of these muscle groups directly.

Previous publications [1] suggest a possible relationship in the close location of the facial motor nucleus and micturition centre at the pons. This may be unlikely as they reside at different locations - the facial motor nucleus lies ventrally in the inferior pons, and the pontine micturition centre (Barrington's nucleus) lies in the dorsal pons at the floor of the fourth ventricle [3].

Disordered control of the anal sphincter is described in two thirds of patients with Ochoa syndrome [1]. Indeed, this symptom is present in one of our patients. The brainstem centre for defecation is yet undefined. However, there is a

common nucleus responsible for control of micturition and defecation at the level of the sacral cord – the Onuf's nucleus, which controls the external urinary and anal sphincters separately through two subnuclei. Careful urodynamic studies may prove whether a disordered innervation and organisation of the Onuf's nucleus explains these symptoms.

CONCLUSION

The neurological mechanisms underlying Ochoa syndrome is intriguing. The presence of an unusual facial grimace when smiling, in association with a neurogenic bladder are clues to the diagnosis. Careful attention to the facial expression in a variety of emotional and voluntary situations may help to solve the reason behind the peculiar facial expressions.

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TRANSPARENCY DECLARATION

Indra Ganesan has nothing to disclose.
Terrence Thomas has nothing to disclose.

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