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Dear Editor,

HYPOGAMMAGLOBULINEMIA IN A 12-YEAR-OLD PATIENT WITH JACOBSEN SYNDROME

Jacobsen syndrome (JS) is a rare congenital disorder with an estimated prevalence of 1 in 100 000 newborns,[1] caused by partial deletion of the long arm of chromosome 11 (del(11)(q23)), which is a *de novo* event in 85% of cases. The main clinical and analytic findings of JS are reviewed elsewhere.[1] The association of JS with an immune alteration (humoral and/or cellular) has been described only sporadically.[2–5]

A boy with JS, currently 12 years of age, is herein described. Diagnosis was established based on clinical findings (thrombocytopenia, umbilical and bilateral inguinal hernia, left hydrocele, right cryptorchidism, severe mental retardation, cardiac alterations, corneal opacity, facial dysmorphism and clinodactyly).

Karyotiping and comparative genomic hybridisation, identified a duplication of the 11q22-q23 region and a deletion in the 11q24.3-qter region. Chromosome study of the parents was normal and the anomaly was considered a *de novo* event.

Since infancy, the patient had presented chronic diarrhoea and multiple respiratory tract infections that required various hospitalisations and antibiotic therapy. At the age of 5, immunologic study demonstrated persistent hypogammaglobulinemia with present but poor response to some vaccines and lymphopenia with no significant cell function alterations (Table 1). Therefore, regular intravenous immunoglobulin (IVIG) therapy was started and the patient experienced an evident clinical improvement with regard to infections. Currently, he remains dependent on his care givers for daily living and he has had no other complications related to the syndrome.

The association of immunologic alterations with JS is uncommon and poorly defined.[2–5] Sirvent *et al*.[2] described two JS patients with a predominantly humoral defect, as in our case, but without an IgG deficit or the need for IG therapy. Von Bubnof *et al*. presented a single adult JS patient with affected cell

**Table 1** Immunological study results

| | Patient results | Normal values* |
|---|---|---|
| IgG | 226–275† mg/dL | (400–1100 mg/dL) |
| IgM | 6–8 mg†/dL | (50–180 mg/dL) |
| IgA | 31–47 mg/dL | (10–160 mg/dL) |
| Total lymphocytes | 896/mm3 | 3300 (2300–5600) |
| T lymphocytes | 80% (717/mm3) | 2300 (1400–3600) |
| CD3 + CD4+ | 51% (457/mm3) | 1300 (700–2000) |
| CD3 + CD8+ | 23% (206/mm3) | 800 (500–1400) |
| CD19+ | 7% (63/mm3) | 860 (400–1500) |
| CD56+ | 8% (72/mm3) | 400 (100–700) |
| Total complement activity (CH50) | 54 UH/mL | (41–90 UH/mL) |
| Cell function study | CD69 + PHA: 90%, | >40% |
| | CD69 + CD4 + anti-CD3: 36%; | >40% |
| | CD69 + CD8 + anti-CD3: 36% | >40% |
| Autoimmune study | ANA, APCA, ASMA, AMA, anti-LKM antibodies, antireticulin antibodies, antitransglutaminase antibodies, antigliadin antibodies and antigliadin IgA: negative | NA |
| Antibody response | Tetanus (0,08 IU/mL), measles (350 mIU/mL) and rubella (14 IU/mL): positive | Tetanus: >0.01 IU/mL |
| | | Measles: >150 mIU/mL |
| | Parotiditis (290): inconclusive | Rubella: >4 IU/mL |
| | *Haemophilus influenza* and *Streptococcus pneumoniae*: negative‡ | Parotiditis: >500 |
| | | Hib and *S. pneumoniae*: Antibody response was considered positive when a 5-fold increase was observed. |
| Antistreptolysin O titer | 200 IU/mL | ≥200 IU/mL |

*Normal values corresponding to the age of the patient at the time of the study. †Immunoglobulin levels in separated samples within 6 months. ‡Hib prevaccination 0,4: postvaccination 0,8 ng/mL and *S. pneumoniae* prevaccination 40: postvaccination → 180 U/mL (total antibody response). Postvaccination titres were evaluated 1 month after vaccination with conjugated HiB (HibTITER) and pneumococcal polysaccharides (Pneumovax 23) vaccines, respectively. AMA, antimitochondrial antibodies; ANA, antinuclear antibodies; APCA, antiparietal cell antibodies; ASMA, antismooth muscle antibodies; CD, cluster of differentiation; LKM, liver-kidney-microsomal antibodies; NA, not applicable.

immunity involving a CD3+CD4+ deficit and decreased proliferative response.[4] Puglisi *et al*. have recently described two cases of 11q terminal deletion disorder and common variable immunodeficiency (CVID).[5]

Although data on the use of IVIG therapy in JS patients is scarce, the clinical and immunological similarities of our case with CVID lead us to assume its effectiveness.

In conclusion, we underline the need for multidisciplinary management of patients with JS and other polymalformative syndromes and execution of an adequate immunological study, particularly if they present severe or repeated infections. The indication to initiate treatment with IVIG should be individualised and determined by the patient's prognosis for survival.

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Dear Editor,

MESENTERIC CYST IN A NEONATE CAUSING OBSTRUCTIVE UROPATHY AND SECONDARY TYPE 1 PSEUDOHYPOALDOSTERONISM – A CASE REPORT

Mesenteric cysts are rare intra-abdominal lesions with an incidence of 1 per 140 000 general hospital admissions and 1 per 20 000 paediatric hospital admissions.[1] Pseudohypoaldosteronism (PHA) is one of the rare conditions in neonates which cause hyperkalaemia and salt loss, accompanied by vomiting, failure to thrive and dehydration.[2] Various presentations of mesenteric cyst have been described in literature.[3,4] An extensive PUBMED search did not reveal a single case of a mesenteric cyst in paediatric age group presenting as secondary type 1 hypoaldosteronism. We herein present a rare case of a mesenteric cyst in a neonate causing obstructive uropathy and secondary type 1 PHA.

In September 2009, a nine-day-old female infant presented with 2 days of refusal to feed, lethargy and anuria. There was no history of fever, convulsions, vomiting or diarrhoea and the infant was exclusively breastfed. She was born at term by spontaneous normal vaginal delivery to a multigravida mother after an unremarkable pregnancy. Ultrasonography was not performed during the pregnancy. She had a birthweight of 3100 g and was discharged on day 3 exclusively on breastfeeding. On examination, the infant was dehydrated, lethargic and afebrile with acidotic breathing. The heart rate was 140/min, respiratory rate, 38/min and mean blood pressure, 38 mmHg. Peripheral pulses were well felt and capillary refill time was 3 s. Her weight was 2200 g indicating a weight loss of about 29%. Abdominal examination revealed a large abdominal mass occupying the whole of the abdomen and descending into the pelvis. It was fixed with ill-defined borders and dull on percussion apart from a small area of resonance due to a part of the overlying bowel. Other systemic examination was normal. The investigations at admission are shown in Table 1. Haemoglobin was 13 g/dL, total leukocyte count was 17 600/cumm and platelet count was 2.6 lac/cumm. Liver function tests, coagulation profile and septic screen including C-reactive protein were normal. The infant was catheterised and 7 mL of urine was recovered. Urinalysis showed protein (1+), occult blood (–), red blood cells 1/high power field, and white blood cells 5/high power field. Urine bacterial cultures were negative. Initial fluid resuscitation was done with 20 cc/kg of normal saline and intravenous cefotaxime was started. This was followed by correction for hyponatremic dehydration. Ultrasonography of the abdomen revealed a well-defined cystic mass measuring $5.8 \times 7.6 \times 5.9$ cm extending inferiorly compressing the ureters and causing pressure effect on both the kidneys leading to bilateral hydronephrosis. The bladder was empty with Foleys catheter tip seen *in situ*. Plain magnetic resonance imaging of the abdomen – T2-weighted images revealed a hyperintense lesion in the presacral space extending inferiorly into the perineum with a few septations within (Fig. 1). Exploratory laparatomy was done on day 10 of life. A huge mesenteric cyst was present in the abdomen between the bladder and the uterus compressing both ureters and causing bilateral hydronephrosis. The cyst was dissected away from the surrounding structures after decompression and a subtotal excision was done. Histopathology of the specimen showed mesenteric lymphangioma with lymph channels and lymphoid aggregates. Postoperatively, the infant was managed in the neonatal intensive care unit. Laboratory parameters done postoperatively serially are shown in Table 1. The infant had gained 256 g on the third day of admission. However, even after adequate correction of dehydration the electrolyte abnormalities and acidosis persisted, and hence secondary type 1 PHA due to obstructive uropathy was suspected. Elevated serum aldosterone and renin levels in the presence of metabolic abnormalities confirmed the diagnosis. Antibiotics and


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