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Pediatric Cardiology

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Protocol for patients with Jacobsen syndrome

Provide this protocol immediately when treating with a healthcare provider unfamiliar with you/your child and Jacobsen syndrome

Relevant Facts:

Jacobsen syndrome (JS) is a rare condition caused by the deletion of the end of the long arm of chromosome 11 (11q).

It is assumed that ALL patients with JS have a specific bleeding disorder called Paris-Trousseau syndrome. Paris-Trousseau has two components:

- 1) Severe thrombocytopenia (low platelet count) of the newborn, which usually returns to near- or low-normal platelet counts during childhood
- 2) Persistent platelet dysfunction

It should be assumed that all patients with JS are at risk for potentially life-threatening bleeding. High risk situations include many kinds of surgeries and trauma.

To healthcare providers: Patients with JS have died when healthcare providers failed to address or were slow to address what were perceived to be low risk bleeding situations. Bleeds, potential bleeds or procedures posing a risk of bleeding should be treated with extreme caution, and serious consideration should be given in such situations to consulting a hematologist and/or an expert in JS.

We have developed the following protocol for the management of bleeding problems in all JS patients:

- Amicar
 - For dental procedures, mouth or nose bleeding
 - Use as a mouthwash or take as a pill or liquid

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Regional Offices:

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- Oral contraceptives
 - For females with heavy periods
- DDAV P
 - May improve platelet aggregation
 - IV or nasal spray
- Platelet transfusion
 - For serious bleeding or before major surgeries

In addition, any medications that impair normal platelet function (e.g., ibuprofen, aspirin) should be avoided.

We recommend the following for patients diagnosed with JS:

Monthly CBC, first 3 months, then once per year.
Platelet function studies, once platelet count normalizes.

If a JS patient is in a life-threatening bleeding situation – or for more information about JS, you may contact Dr. Paul Grossfeld 24/7 at 858-576-1700. Please ask the page operator to page Dr. Grossfeld. If Dr. Grossfeld is unavailable please ask the page operator to contact Dr. Amy Geddis (pediatric hematologist). If neither Dr. Grossfeld nor Dr. Geddis are available, please ask the page operator to contact the on-call pediatric hematologist.

This protocol is based on current understanding of Paris-Trousseau and JS at a center with significant experience with this problem. Our protocol is subject to change as further information is obtained in the future. Our most current JS protocol is always available at www.11qusa.org.

Sincerely Yours,



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