Fechtner Syndrome—A Variant of Alport's Syndrome With Leukocyte Inclusions and Macrothrombocytopenia

By LoAnn C. Peterson, K. Venkateswara Rao, John T. Crosson, and James G. White

This study reports a family comprising four generations in whom nephritis, deafness, congenital cataracts, macrothrombocytopenia, and leukocyte inclusions were observed in varying combinations in eight of 17 members. The family differs from others reported in that their hematologic abnormalities include not only macrothrombocytopenia, but also small, pale blue cytoplasmic inclusions in the neutrophils and eosinophils. Light microscopic appearance of the inclusions resembled that of toxic Döhle bodies and inclusions of May-Hegglin anomaly, but their ultrastructural appearance was unique. The inclusions consisted of clusters of ribosomes and small segments of rough endoplasmic reticulum (RER). They lacked the parallel 10-nm filaments characteristic of May-Hegglin anomaly and the parallel strands of RER seen in toxic Döhle bodies. Platelets were large, but their light and ultrastructural appearance was not significantly different from normal platelets. Platelet aggregation in response to epinephrine, arachidonate, thrombin, adenosine diphosphate, collagen, and ristocetin was normal. Levels of nucleotides and serotonin were elevated in proportion to cell volume. The concentration of adenosine triphosphate secreted and the percentage of arachidonate acid converted to thromboxane B₂ were proportional to cell number. Deafness was high-tone sensorineural. Renal disease ranged from microscopic hematuria to end-stage renal failure necessitating dialysis and kidney transplantation. All affected adults had cataracts. This family represents a variant of Alport's syndrome with cataracts and leukocyte inclusions that, because of the associated macrothrombocytopenia, may be confused with May-Hegglin anomaly.

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The propositus (B2) is a 39-year-old Caucasian male who has suffered from epistaxis and easy bruising since childhood. At age 18, during a physical examination for induction into the army, he was found to be hypertensive and to have albuminuria. Subsequent studies showed a normal blood urea nitrogen (BUN) and a normal intravenous pyelogram. He was treated for his hypertension, but his renal function progressively deteriorated, necessitating dialysis at age 23. After one month of dialysis, he underwent bilateral nephrectomy and splenectomy in preparation for a cadaver renal allograft, which he received in June 1972. The pathology of the nephrectomy specimen is given below. The transplanted kidney was removed after six weeks because of irreversible graft rejection and the patient remained on dialysis until August 1976, when he received a second cadaver renal transplant. During the interval, the patient had two bleeding episodes, one a spontaneous hematoma of the rectus abdominus muscle and the other an upper gastrointestinal bleed secondary to gastritis. The second renal allograft has functioned well, and the patient is employed full-time by a local electronics firm.

The physical examination was normal except for obesity and surgical scars. The blood pressure was 130/80 mmHg. Slit lamp examination of the eyes revealed bilateral cerulean congenital cataracts. Audiometric evaluation showed bilateral moderate high-tone sensorineural hearing loss with maximum loss of 80 dB observed at a frequency of 8,000 Hz (cps).

Routine laboratory testing gave the following results: hemoglobin 13.2 g/dl, white blood cell count 11.3 × 10⁹/L with normal differential, platelet count 96 × 10⁹/L. Microscopic examination of the peripheral blood smear showed giant platelets and cytoplasmic inclusions in the neutrophils and eosinophils (described in detail below). The bleeding time, prothrombin time (PT), activated partial thromboplastin time (aPTT), thrombin time (TT), and fibrinogen level were normal. Urinalysis was normal. BUN was 15 mg/dL, serum creatinine was 0.9 mg/dL, and creatinine clearance was 120 mL/min. Serum electrolytes, calcium, phosphorus, albumin, and lipids were normal. Serum and urine osmolalities obtained after an overnight fast were 284 and 501 mosm/kg, respectively.

Family History

The family pedigree involving four generations is shown in Fig 1. Histories were obtained and physical examinations were performed on the four siblings (B1, B3, B4, B5) and the niece (C2) of the propositus by one of us (K.V.R.). Audiometric, ophthalmic, and renal evaluations were also carried out. Blood specimens were obtained from all 16 living members of the family.

The father (A1) died at age 69 after diabetic gangrene and sepsis. A peripheral blood smear that was obtained while the patient was alive showed macrothrombocytopenia and leukocyte inclusions simi-
Thrombocytopenia leukocyte inclusions; t3 I!. high tone deafness; 11. macro- 
volumes were analyzed on platelet-rich plasma (PRP) in an elec-
ted to the propositus. There was no history of bleeding tendency, 
renal disease, or hearing or visual problems.

Two of the siblings (B1 and B4) had macrothrombocytopenia and 
leukocyte inclusions. The platelet counts were 54 x 10^9/L (B1) and 
40 x 10^9/L (B4). Both had normal coagulation tests, including 
bleeding time, PT, aPTT, TT, and fibrinogen level. The sister, age 
40, had a history of easy bruising, prolonged bleeding after tooth 
extractions, and uterine bleeding, which necessitated hysterectomy 
at age 29. She also had a bleeding peptic ulcer at age 38. The 
brother, age 34, gave a history of easy bruising since childhood. Both 
of the affected siblings had bilateral high-tone hearing loss. Both had 
microscopic hematuria (4 to 6 rbc/hpl), but the renal function tests 
were normal, including BUN, creatinine clearance, serum electro-
lytes, and serum osmolality. The sister’s ophthalmic examination 
was normal; the brother had cerulean cataracts. The younger sister 
(B3), age 37, had congenital cataracts but no other abnormalities 
associated with this syndrome. The youngest brother of the patient 
(B5), age 28, had conductive deafness, perforated tympanic mem-
branes, cerulean cataracts, and a seizure disorder of unknown 
etiology but no hematologic abnormalities.

Only one person from the third generation was affected with this 
disorder. This 17-year-old female (C2) had macrothrombocytopenia 
(platelet count 30 x 10^9/L) and leukocyte inclusions. She also had 
microscopic hematuria, recurrent urinary tract infections, and con-
genital cataracts. There was no history of deafness or a bleeding 
tendency.

Her son (D3), age 18 months, also had macrothrombocytopenia 
(platelet count 120 x 10^9/L) and leukocyte inclusions. There was no 
history of bleeding tendency, deafness, or renal disease.

**MATERIALS AND METHODS**

**Light Microscopy and Cytochemistry**

Blood smears were stained with Wright’s-Giemsa, periodic acid 
Schiff (PAS), nonspecific esterase, myeloperoxidase, acid phos-
phatase, and methyl green pyronin. Leukocyte alkaline phospha-
tase scores were determined on all affected living adult patients.

**Platelet Counts**

Blood was obtained by venipuncture into a vacutainer tube with 
EDTA as the anticoagulant. The sample was diluted in a Unopette 
(Becton Dickinson, Rutherford, NJ), placed on a hemocytometer, 
and counted in duplicate by phase microscopy. From the number of 
cells seen, the total platelet count was calculated.

**Platelet Volume**

Blood for platelet sizing was collected from four affected individu-
als into a vacutainer tube containing EDTA, and another sample was 
collected in a vacutainer containing trisodium citrate. Platelet 
volumes were analyzed on platelet-rich plasma (PRP) in an elec-
tronnic particle sizing system (Coulter Counter model ZB and 
Channelizer C-1000, Hialeah, Fla) as described previously.

**Platelet Function**

Platelet function studies were performed on all affected 
adults. After venesection, the sample was mixed immediately with 
citrate-citric acid dextrose (93 mmol/L sodium citrate, 7 mmol/L 
citric acid, and 140 mmol/L dextrose), pH 6.5, in a ratio of nine 
parts blood to one part anticoagulant. PRP was separated from 
whole blood by centrifugation at room temperature for 20 minutes at 
100 g. Platelet aggregation studies were performed using a Payton 
(Buffalo, NY) dual channel aggregometer with PRP and platelet-
poor plasma. Aggregants added to PRP included acid-soluble collagen 
(Worthington, Freehold, NJ) at 30 to 100 μg/mL, epinephrine 
at 5.5 to 100 μmol/L, bovine thrombin (Parke-Davis, Detroit) at 0.1 
to 0.4 U/mL, the sodium salt of arachidonic acid (greater than 99% 
pure, Nu Chek Prep, Elysian, Minn) at 0.45 to 0.9 mmol/L, and 
ristocetin (Helena, Beaumont, Tex) at 1.5 mg/mL.

A Lumiaggregometer (Chronolog Corp, Havertown, Pa) was used to 
study the aggregation and release reaction simultaneously. 
Luciferase (4 μg/mL) was added to each platelet sample just before 
the stirring bar on the aggregometer. Chemiluminescence and 
aggregation were recorded by upward deflections of their recording 
pegs on moving graph paper. Each response was calibrated by 
addition of adenosine triphosphate (ATP) at a final concentration of 
4 x 10^-4 mol/L, and the approximate amount of secreted ATP 
estimated by the fractional relationship of upward deflections caused 
by release and by addition of standard times the known concentra-
tion of ATP.

**Platelet Biochemistry**

Platelet biochemical studies were performed on platelets from 
the propositus and his two affected siblings. Nucleotide levels were 
quantitated by high-pressure liquid chromatography according to 
the procedure developed in this laboratory. Serotonin was 
extracted and measured by the method of Rao et al. The conversion 
of arachidonic acid by platelet cyclooxygenase was evaluated by a 
modification of the method of Hamburg and Samuelsson using 
14C-arachidonic acid (Applied Sciences, State College, Pa) as sub-
strate.

**Ultrastructural Studies of Platelets and Leukocytes**

Samples of PRP and buffy coats were combined with equal 
volumes of 0.1% glutaraldehyde in White’s saline, pH 7.3 (a 10% 
solution of a 1:1 mixture of (a) 2.4 mol/L NaClO, 0.1 mol/L KCl, 46
Table 1. Platelet and Neutrophil Studies of Living Adult Family Members With Leukocyte Inclusions and Macrothrombocytopenia

<table>
<thead>
<tr>
<th>Patient Characteristics and Tests Done</th>
<th>Pedigree Designation*</th>
<th>Control Subjects</th>
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<tbody>
<tr>
<td>Age (yr)</td>
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<tr>
<td>Sex</td>
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<td>Platelet count x 10⁹/L</td>
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</tr>
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<tr>
<td>Ristocetin</td>
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<td>+</td>
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<td>Neutrophil function (chemiluminescence assay)</td>
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</table>

N, normal as compared with matched control; +, present; ND, not done; Δ, proband.

* Pedigree designation is as given in Fig 1.

mmol/L MgSO₄, 64 mmol/L Ca (NO₃)₂ 24 H₂O; and (b) 0.13 mol/L NaHCO₃, 8.4 mmol/L NaHPO₄ · 1 H₂O, 3.8 mmol/L anhydrous KH₂PO₄, and 0.1 g/L phenol red). After 15 minutes at 37 °C, the samples were sedimented to pellets and the supernatant was discarded and replaced with 3% glutaraldehyde in the same buffer. Fixation was continued at 4 °C for 60 minutes. The cells were then washed in buffer and combined with 1% osmic acid in veronal acetate (0.02 N HCl and a 20% solution of a stock buffer solution containing 0.14 mol/L sodium barbital, 0.145 mol/L sodium acetate · 3 H₂O, and a 6.8% solution of a stock salt solution containing 1.7 mol/L NaCl, 54 mmol/L KCl, and 18 mmol/L CaCl₂). After exposure to the second fixation for one hour, the cells were dehydrated in a graded series of alcohol and embedded in Epon 812. Contrast of thin sections cut from plastic blocks on an ultramicroscope was enhanced with uranyl acetate and lead citrate. Observations were made in a Philips 301 electron microscope. The results were compared with 12 patients with May-Hegglin anomaly previously studied in the same manner by one of the authors (J.G.W.).

Fig 3. Thin section of buffy coat sample from peripheral blood of a patient with the Fechtner syndrome. Many giant platelets, some larger than the two lymphocytes (L) are apparent in the sample (original magnification ×5,000; current magnification ×4,000).
Functional Evaluation of Neutrophils

The luminol-enhanced micro chemiluminescence assay was used to study the oxidative metabolic responses of the neutrophils of the propositus as compared with a normal control. The chemiluminescence mixtures in glass scintillation vials consisted of $2.5 \times 10^8$ phagocytic cells, $1 \mu$m luminol (Eastman Kodak Co, Rochester, NY), and 5 mg of preopsonized zymosan (Sigma Chemical Co, St Louis) or $1 \text{nmol/L}$ phorbol myristate acetate (PMA) (Sigma) in a final volume of 5.5 mL of Hanks' balanced salt solution containing 0.1% gelatin. Before adding the stimuli (zymosan or PMA), background counts of phagocytes alone were obtained. After addition of the stimuli, counts were again obtained every three minutes for 30 minutes and expressed as the number of counts per minute per $10^9$ cells.

Renal Pathology

Two-micrometer sections were cut from the nephrectomy specimen and stained with hematoxylin and eosin, trichrome, and silver stains. Ultrastructural studies were performed on formalin-fixed paraffin-embedded tissue.

RESULTS

Platelets

Light microscopy. Giant platelets were seen in the peripheral blood smears of all affected family members (Fig 2). There was marked variation in the size of the platelets in each patient, but many of the platelets were larger than erythrocytes and some were larger than lymphocytes.

Volume. The mean platelet volumes of three affected patients were increased, as shown in Table 1. There was no significant difference in the size of the platelets whether EDTA or citrate was used as the anticoagulant, and the sizes reported are those obtained when the citrate was the anticoagulant. Because some of the larger platelets were probably sedimented with theuffy coat, these measured volumes may underestimate the actual mean platelet volumes.

Ultrastructural studies. Thin sections of the platelets from affected family members revealed the wide variations in platelet size evident on peripheral smears (Fig 3). The platelets that were near normal in size were discoid and supported by circumferential bundles of microtubules. Most of the giant platelets were spherical (Fig 4). Bands and bundles of microtubules were apparent under the cell membranes but were seldom organized in a single plane as in discoid platelets. Granules, dense bodies, and occasional mitochondria were randomly dispersed in the cytoplasm. Glycogen in the form of single particles and masses was evenly deposited in the cytoplasm of the giant platelets. Elements of the surface-connected open canalicular system and channels of the dense tubular system were spread evenly within the large cells. Large membrane complexes formed by the intertwining of elements from the two membrane systems were commonly observed. Except for the large size and relatively spherical form, the ultrastructural appearance of these platelets was not significantly different from that of normal controls.

Functional studies. Samples of PRP from the affected family members responded in the same manner as normal platelets when stirred with aggregating agents on the platelet aggregometer (Table 1). The tracings differed because the patient samples contained giant platelets at concentrations of 50 to $100 \times 10^9/L$, whereas control samples had normal-sized platelets at counts of $300 \times 10^9/L$. The platelets from the patients responded biphasically to epinephrine and irreversibly to concentrations of arachidonate, thrombin, ADP, collagen, and ristocetin that caused irreversible aggregation in stirred samples of normal PRP.

Fig 4. Giant platelet from another patient with Fechtner syndrome. Although the cell is large, the relative numbers of granules (G), mitochondria (M), and dense bodies (DB) is not unusual. Microtubules (MT) and elements of the dense tubular system (DTS) of channels are present (original magnification $\times 26,500$; current magnification $\times 21,730$).
Simultaneous aggregation and release of ATP from platelets of three affected family members (B2, B4, C2) was observed in a platelet Lumiaggregometer (Fig 5). The aggregation response was similar to that of normal platelets. Amounts of ATP released by the giant platelets were generally less than secreted by control cells exposed to the same concentrations of aggregating agents. The amounts of ATP released from the large cells, however, were sufficient to support irreversible aggregation.

**Biochemistry.** Concentrations of adenine and guanine nucleotides were significantly increased in Fechtner syndrome platelets, but their ATP-ADP ratio was identical to that found in normal platelets (Table 1). Serotonin concentrations in platelets were also increased (Table 1). The ability of the giant platelets to convert $^{14}$C-arachidonic acid into thromboxane B$_2$ appeared reduced to about one third that of normal platelets (Table 1).

**Leukocytes**

**Light microscopy.** The neutrophils of all affected patients contained one to several small, 1- to 2-μm, irregularly shaped, cytoplasmic inclusions that appeared pale blue with Wright’s-Giemsa stain (Fig 6). They were present in most neutrophils and occasionally were observed in eosinophils. The inclusions closely resembled the spindle-shaped inclusions seen in May-Hegglin anomaly, but they were smaller and stained less well. They differed from Döhle bodies associated with septicemia, since they were present in most cells and were not associated with any other toxic morphologic changes.

The neutrophils stained normally with PAS, nonspecific esterase, myeloperoxidase, and acid phosphatase. There was faint positive staining of the cytoplasmic inclusions with methyl green pyronin. Leukocyte alkaline phosphatase scores were normal in all affected patients except for one (C2), who had an elevated score concurrent with a urinary tract infection.

**Ultrastructural studies.** Thin sections of neutrophils from affected members of the Fechtner family resembled those from normal individuals. The only clear difference was the presence of an unusual inclusion in the cytoplasm of the Fechtner neutrophils (Fig 7), which differed in appearance from other types of neutrophil inclusions. Fechtner inclusions might be mistaken for the May-Hegglin anomaly. However, Fechtner inclusions were small and irregular (Fig 7), and the May-Hegglin inclusions, large and spindle-shaped (Fig 8). A linear array of parallel 7- to 10-nm filaments was oriented in the long axis of the spindle-shaped May-Hegglin inclusions (Figs 8 and 9A, B). The Fechtner inclusions consisted of zones of cytoplasm free of granules, glycogen particles, and other organelles (Figs 7 and 10, A and B). Like the May-Hegglin anomaly, the Fechtner inclusions contained clusters of single ribosomes in addition to small segments of rough endoplasmic reticulum, and they were not isolated from surrounding cytoplasm by an enclosing membrane (Fig 10A, B). The major difference between the May-Hegglin anomaly and the Fechtner inclusions was the absence of parallel 7- to 10-nm filaments from the latter structures (Figs 9, A and B, and 10, A and B).

Fechtner inclusions might also be confused with the Döhle bodies found in neutrophils associated with septicemia, especially when studied with the light microscope. At the ultrastructural level, however, there was no similarity. Toxic Döhle bodies consist of segments of rough endoplasmic reticulum arranged in a parallel fashion (Figs 11 and 12). Similar structures were not seen in neutrophils from the Fechtner family or in neutrophils from patients with the May-Hegglin anomaly.

**Neutrophil function studies.** The chemiluminescence response of the patient’s neutrophils to opsonized zymosan (725 cpm/10$^3$ cells) and PMA (740 cpm/10$^3$ cells) was normal as compared with responses observed from neutrophils of normal controls (515 cpm/10$^3$ and 535 cpm/10$^3$ cells, respectively).
Renal Histology

The sections obtained from the nephrectomy specimen showed an end-stage kidney. The majority of the glomeruli were hyalinized; however, the preserved glomeruli were hypercellular with increased mesangial cells and matrix. There was patchy tubular drop out. The remaining tubules appeared dilated with an eosinophilic cast-like material in the lumen. The vessels demonstrated moderate medial thickening. A diffuse interstitial cellular infiltrate consisting of small mononuclear cells and a few plasma cells was present. The degree of interstitial cellular infiltrate was greater than would be expected for this stage of renal destruction and indicated an interstitial disease paralleling the glomerular sclerotic process. These features are suggestive of hereditary nephritis.

Ultrastructurally, glomerular basement membranes were thickened and tortuous with focal areas of attenuation, a finding also suggestive of hereditary nephritis. However, the lamina densa showed no significant alterations. There was focal fusion of epithelial podocytes. The mesangium was unremarkable. There were no electron-dense deposits either in the mesangium or along the capillary wall. Tubular basement membranes were normal. Focal interstitial fibrosis was present. These morphologic findings are consistent with the diagnosis of hereditary nephritis.

DISCUSSION

The present report describes a family in which eight individuals from four generations have a constellation of inherited abnormalities not previously reported.
Inclusion bodies in the cytoplasm of neutrophils from a patient with the May-Hegglin anomaly. The inclusions are spindle-shaped structures, but may appear round or oval in thin sections. Small segments of rough endoplasmic reticulum (ER) are present at the edge and sometimes within the inclusions, but they are not bounded by a membrane. The matrix is less electron dense than the cytoplasm and is virtually free of granules (G). Ribosomes (R), smaller and less dense than glycogen particles (Gly), are present singly and in clusters. The distinguishing feature of the inclusion are the 7 to 10 nm filaments (F) lying parallel to each other in the long axis of the spindle-shaped structures (9A original magnification x46,000, current magnification x38,880; 9B original magnification x50,000, current magnification x39,000).

Characteristic clinical features include macrothrombocytopenia, leukocyte inclusions, hereditary nephritis, sensorineural deafness, and cataracts. We have called this unusual association of findings the Fechtner syndrome after the last name of the propositus.

The critical feature of the Fechtner syndrome separating it from other disorders of a similar type is the presence of cytoplasmic inclusions in many circulating neutrophils and some eosinophils. The inclusions resemble the inclusions found in granulocytes and

Inclusions in the cytoplasm of neutrophils from a patient with Fechtner syndrome. Fechtner inclusions were more difficult to find in thin sections than May-Hegglin inclusions because they were small and not spindle-shaped. The irregular inclusions contain bits of rough endoplasmic reticulum (ER) and are devoid of granules. Numerous ribosomes (R), usually in clusters, are present in the matrix which is not demarcated by a membrane. The parallel 7- to 10-nm filaments of the May-Hegglin anomaly are absent in Fechtner inclusions (10A original magnification x36,000, current magnification x28,080; 10B original magnification x50,000, current magnification x39,000).
monocytes of patients with the May-Hegglin anomaly, but they are smaller in size and less well stained. At the ultrastructural level, the difference is striking. Fechtner inclusions contain clusters of ribosomes and fragments of rough endoplasmic reticulum but lack the parallel bundles of 10-nm filaments characteristic of inclusions in May-Hegglin granulocytes.

By light microscopy, the Fechtner inclusions are similar to Döhle bodies found occasionally in neutrophils of patients with septicemia. The Fechtner inclusions differ in that they are present in nearly every cell and the cells lack other toxic morphologic features. Ultrastructurally, the parallel orientation of rough endoplasmic reticulum present in Döhle bodies was not observed in the Fechtner inclusions.

Hereditary nephritis is often associated with other inherited defects. Congenital deafness appears to be the most common, and the two abnormalities occurring together constitute Alport's syndrome. In 1960, Epstein et al reported two families in which giant platelets were associated with hereditary nephritis and deafness. Function of the giant platelets was found to
be abnormal. Subsequently, Eckstein et al² described a family with macrothrombocytopenia, deafness, and renal disease, but platelet function in the affected members was normal. Granulocyte inclusions were not observed in either the Epstein syndrome or the family reported by Eckstein.

Brivet et al⁷ have reported a family with the May-Hegglin anomaly and hereditary nephritis. None of the members in this family has deafness or cataracts. Detailed morphologic, cytochemical, and ultrastructural studies on the granulocyte inclusions were not performed, but there seemed to be no question that the cytoplasmic inclusions were consistent with the May-Hegglin anomaly in affected family members.

It is not possible to state with absolute certainty that Brivet’s cases⁷ did not have the same syndrome as the Fechtner family. However, the absence of cataracts and deafness, together with what was characterized as the May-Hegglin anomaly, suggest that this form of hereditary nephritis differs from the Fechtner syndrome presented here. Thus, the Fechtner family appears to constitute a novel variant of hereditary nephritis and to present a new type of granulocyte inclusion.

Giant platelets from members of the Fechtner family were huge. Many of them were larger than lymphocytes and a few exceeded the size of neutrophils, eosinophils, and monocytes. Despite their massive size, the giant platelets were not significantly different from normal platelets in their morphologic features.¹¹ All contained granules, dense bodies, mitochondria, glycogen, and elements of the open canalicular system and dense tubular system. The number of organelles per cell was increased in proportion to the expanded cell volume, but the density and distribution did not differ from that observed in the cytoplasm of normal platelets. The only unusual feature was the prominence of channels of the open canalicular system and the membrane complexes they form with elements of the dense tubular system. In this respect, Fechtner platelets are essentially identical to cells from patients with the May-Hegglin anomaly¹² and Epstein’s syndrome.¹

The function and biochemistry of Fechtner platelets were normal. Concentrations of aggregating agents causing irreversible aggregation in control PRP produced similar responses in Fechtner platelet samples. Levels of nucleotides and serotonin were elevated in Fechtner platelets, but the increase was in proportion to the expanded cell volume. ATP-ADP ratios were similar to normal platelets. Although the amounts of chemical constituents in each cell were increased compared with normal platelets, the concentration of ATP secreted after activation and the percentage of arachidonic acid converted to thromboxane B₂ were more proportional to cell number than to cell mass. The basis for this apparent discrepancy is unknown but did not appear to affect irreversible aggregation of Fechtner platelets.

Renal disease in the Fechtner family appears similar to that in patients with classic hereditary nephritis.⁸ The propositus presented with albuminuria and progressed to end-stage renal failure, necessitating dialysis and renal transplantation. Even though the kidney was end stage at the time of the transplant nephrectomy, there were morphologic features that supported the diagnosis of hereditary nephritis and that were indistinguishable from those reported in Alport’s syndrome. Two siblings and one niece had microscopic hematuria. Deafness in Fechtner family members was also similar to that reported in families with hereditary nephritis, with or without macrothrombocytopenia ¹⁴. Cataracts identified in our patients resembled those observed in patients with Alport’s syndrome, but this finding has not been reported in association with macrothrombocytopenia.

The present report has described a family with the unusual clinical findings of hereditary nephritis, congenital deafness, cataracts, macrothrombocytopenia, and inclusions in circulating leukocytes. No pathophysiologic explanation has been advanced to explain the many associated phenomenon that occur in patients with hereditary nephritis. Continued basic studies of fundamental defects in the different cells and tissues involved may reveal the common abnormality that links them together.

REFERENCES

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Fechtner syndrome--a variant of Alport's syndrome with leukocyte inclusions and macrothrombocytopenia

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