DDC clinic
Center for Special Needs Children

Fall-Winter 2017-2018
Chronicle

Enhancing the quality of life for people with special needs caused by rare genetic disorders.

Communities Come Together!

The 2017 auction season was a major success thanks to the communities of Shiloh, Spartansburg and Geauga.

Each year, friends in several Amish and Mennonite communities hold benefit auctions in support of our work with special children. Many hundreds of local community members participate, and their commitment to our mission and our families is truly humbling. Volunteers make up the committees with numerous other hands helping to set up, tear down and work during the auction. We greatly appreciate the hard work of the many hands and the generous support of the communities. 2017 was another record setting year!

Shiloh Auction

This was the eleventh year that our clinic has shared in the proceeds from the Shiloh Mennonite Community Benefit Auction. A record number of participants attended the July 8th event. This annual auction benefits both DDC Clinic and the Clinic for Special Children in Lancaster, Pennsylvania. This year's proceeds for DDC Clinic were $40,700 and we are so grateful to this community! Next year's date is July 14, 2018 and we hope to see you there.

Spartansburg Auction

On the 5th of October, the 5th Annual Spartansburg Auction benefit was held in

> PAGE 4

Clinic Research Furthers Understanding of Genetic Risk Factors for Stroke

We are pleased to report the first successful reversal of cerebral blood vessel narrowing in pediatric patients at risk for early onset stroke.

Increasing evidence shows that there is a genetic predisposition to cerebrovascular disorders and that these risk factors may account for a significant portion of unexplained strokes.

In 2011, we first reported on a new genetic condition, SAMS Association, which causes cerebral vasculapathy, a disease that narrows the brain's blood vessels. Our discovery of the condition was published in the *Proceedings of the National Academy of Sciences*.

> PAGE 3

About DDC Clinic

DDC Clinic – Center for Special Needs Children is a non-profit primary care and research facility serving patients with complex medical needs. Located in Middlefield, Ohio, we have been recognized internationally for our state-of-the-art genetic research impacting broad-based health concerns such as heart disease, cancer and neurological disorders.

Our mission is to enhance the quality of life for people with special needs caused by rare genetic disorders. Founded in 1998 by five local Amish families committed to helping tomorrow's special children, we now serve more than 900 patient families in 30 U.S. states and several foreign countries.

Our clinic is a unique collaboration of the Amish and non-Amish communities, dedicated doctors and researchers all working together to find answers for parents and treatments for children. The Clinic was built on the 'medical home' concept defined by the American Academy of Pediatrics. We provide comprehensive and personalized medical care for special children with over 70 different rare conditions.

Our in-house certified clinical genetics laboratory provides rapid, high quality and affordable testing and we conduct patient centered research – always seeking faster diagnoses, better understanding and improved treatments for our children.

We offer personalized education and support services for patient families. No family is ever turned away based on ability to pay.

DDC Clinic is a gathering place; a place of love, compassion and caring; a place where people take the time to listen and share; a place of faith and hope.

7th International Cohen Syndrome Family Gathering

As awareness of Cohen Syndrome and diagnosis rates continue to rise, it is important for families to seek information from the professionals.

With over 40 cases of Cohen Syndrome in our community, DDC Clinic and Dr. Wang have become essential to the needs of families all over the world. What began in 2004 with a small group of families in the local community has grown to an association with hundreds of members worldwide.

The Cohen Syndrome Association is very excited to announce the 2018 location for the next CSA conference. The group is growing in numbers and making an impact globally. This year's venue will provide the needed space for their families. The conference will be held June 21-23, 2018 at The Embassy Suites in Beachwood, Ohio. Registration and more information for the event is available on the Cohen Syndrome Association website, http://cohensyndrome.org.

Congratulations!

DDC Clinic's nurse, Valerie Sency, has been chosen as a finalist for a 2017 March of Dimes Ohio Nurse of the Year in the Pediatric category! Over 400 nurses were nominated across 18 categories representing over 50 health systems and universities. Congratulations, Valerie, on being recognized as a nurse leader!

Clinic Research Furthers Understanding of Genetic Risk Factors for Stroke > from page 1

This condition is an auto-inflammatory syndrome and there is no accepted therapy to successfully prevent disease progression. Brain MRIs of affected children often reveal multiple sites of arterial narrowing - putting them at major risk for aneurysm, stroke and severe functional and cognitive debilitation.

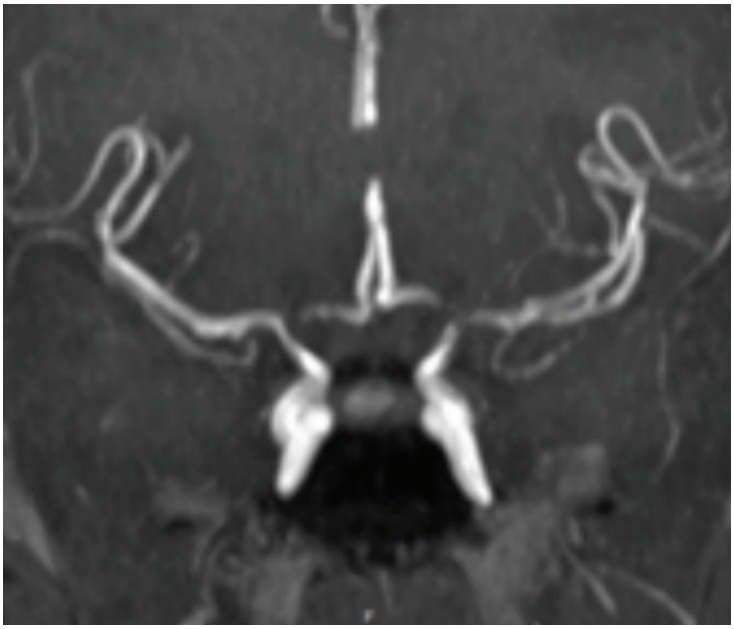
This rare genetic disorder was dually named for the mutated SAMHD1 gene, and as an acronym for the characteristics of the cluster of cerebrovascular problems it causes: *Stenosis, Aneurysm, Moyamoya and Stroke*. In addition to the highly specific narrowing of larger brain arteries, affected patients also have poor blood circulation in their limbs, reduced flexibility in their joints, low muscle tone, short stature, irritability and a number of abnormal laboratory findings. Our clinic now sees more than twenty patients affected by this SAMHD1 variant and consults with clinicians and researchers treating patients at Harvard Medical School, University of Pennsylvania and as far as Israel.

In a 2013 newsletter, we shared with you our initiation of a research collaboration with Case Western Reserve University and Cincinnati Children's Hospital. This collaboration included the launch of a new clinical trial involving the administration of anti-inflammatory infusions to help reduce the risk of pediatric stroke in patients affected by the SAMHD1 mutation. Prior to this trial, therapeutic options for families of affected children were limited. Steroid therapeutics (prednisone) helped with symptoms caused by inflammation, but not the arterial narrowing and the risk for stroke.

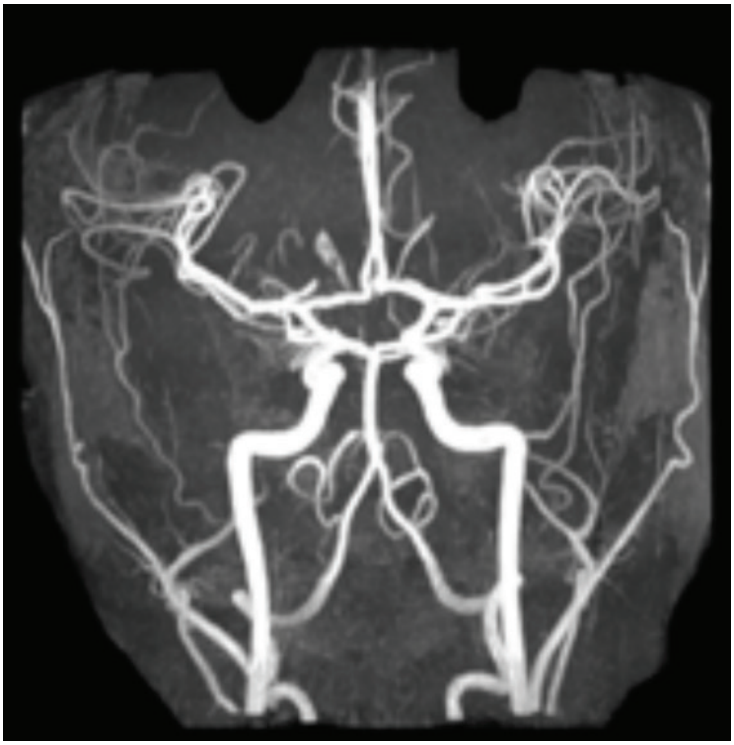
DDC Clinic's research into the impaired function of this specific SAMHD1 mutation has allowed us to work with collaborators to identify other possible and more targeted therapeutic agents. In this case, our growing understanding of the molecular biology led us to design a trial utilizing an anti-inflammatory agent (tocilizumab) developed to help control rheumatoid arthritis.

Over the last four years, six patients have been receiving regular treatments at DDC Clinic and the results have been significant. We are pleased to report the first successful reversal of cerebral blood vessel narrowing in pediatric patients at risk for early onset stroke! One patient obtained disease control within two months and substantial reversal of his cerebral arterial narrowing after one year of treatment. Several other patients, the youngest in the cohort, have also seen disease control and reversal over the last several years. In addition to significantly reducing the risk of stroke, patients in the trial have seen much-improved joint mobility and developmental assessments have demonstrated improved cognition as well. With what we have learned to date, further research is in order to demonstrate that tocilizumab may offer a means in controlling cerebral vasculapathy and other symptoms in children affected by SAMS.

Earlier this year, we shared the results of our ongoing research and this trial in a peer-reviewed publication and have since had inquiries from clinicians with affected patients in New Jersey, Illinois, Massachusetts and Israel.



Patient brain MRI prior to treatment



Patient brain MRI after 2 months treatment.

"Our work continues to bring hope to families both within and outside our community. DDC Clinic's founding board members had the vision that our clinic be a center of excellence for research and provide families highly personalized medical care. Through our ongoing research, clinical trials and publications, we are pleased to share what we have learned so that the work we do in partnership with the local Amish community can benefit other children and families across the country and the world" Dr. Heng Wang, Medical Director.

Many Thanks to...

DDC Clinic is grateful for the support from the following friends:

Ohio Developmental Disabilities Council
For the $50,000 grant award in continued support of targeted research and community outreach services.

United Way Services of Geauga County
Thank you for your support of the Preventative Personalized Medicine Program with a grant in the amount of $20,000. We also wish to recognize the Youth Fund Distribution Committee for a grant of $3,000 for our Patient Assistance Program.

Bessie Benner Metzenbaum Fund
We are grateful for the fund's ongoing commitment to our mission and program services. Thank you for the general operating support award of $10,000.

The Ortino Family Foundation
For their longtime support and most recent gift of $5,000.

William & Margaret Clark Charitable Gift Fund
For their continued support of the clinic and $3,000 gift in operating support.

The Children's Guild
Thank you to the Children's Guild for their commitment to our work and their most recent award of $2,000 in general operating support.

Christ Child Society
Thank you to the Christ Child Society for their continued support and their award of $2,000 for our Patient Assistance and Nutrition Program.

In support of retaining a new physician-scientist who will provide patient care and lead complex research projects and clinical trials, we established the Second Physician Scientist Fund. A special thanks to these visionary donors.

The Catherine L. and Edward A. Lozick Foundation
For the $180,000 three-year challenge grant.

The Cleveland Foundation
For the $135,000 two-year grant award.

Communities Come Together! > from page 1

Crawford County in Western Pennsylvania. A number of families from the region travel to our clinic for their children's care. Several years ago the community came together wanting to help support our mission. We are humbled by the generosity of this small community raising $33,000 at this year's event! Join us next year on August 10, 2018.

Geauga Auction

The 17th Annual Geauga Benefit Auction was held on Saturday, October 14th, and was a success! Beginning at 5:30am with breakfast for those early risers, a hearty meal was provided. New donations to the auction this year were a 16' x 20' timber frame pavilion and a day's labor by various DDC Clinic board members. At the end of the day, more than $180,000 was raised for DDC Clinic! We are humbled by the support of so many. We hope that you will make an effort to join us on October 13, 2018!

If you've never been to one of our traditional benefit auctions, please consider doing so and bring your family & friends!

Patchwork 2017 BENEFIT

We'd like to extend our sincere appreciation to those who attended our annual Patchwork Benefit on Saturday, April 8th.

DDC Clinic – Center for Special Needs Children welcomed over 240 people to enjoy a delightful evening in support of a worthy cause when it hosted its 17th annual Patchwork Benefit on Saturday, April 8th. Proceeds of $91,000 were raised from the event, helping to support the clinic's mission to enhance the quality of life for people with special needs caused by rare genetic disorders.

Highlights of the evening included a selection of hymns sung by our Amish friends. Many described the singing as inspiring and beautiful. A special program presented by Dr. Heng Wang, DDC Clinic Medical Director, followed. Dr. Wang spoke about the first patient he saw at the newly established clinic in 2002. He told guests of the journey of Rebekah and her family. He revealed that in 2016, after many years of not having a diagnosis, he and the staff at DDC Clinic finally had an answer for her family. Rebekah's father also spoke of their journey and how having a diagnosis has changed things for his daughter.

"The Patchwork Benefit is the coming together of Amish and non-Amish communities, and the many partners and supporters that helped us develop a special place for those with complex special needs," said Blake Andres, Executive Director of DDC Clinic.

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COMMUNITY PROGRAMS AND SERVICES

Community Groups

DDC CLINIC QUILTING AND CRAFT CLUB continues to meet once a month. The group has continued to grow since beginning in 2011 with just a handful of ladies. On the first Wednesday of the month from 9:30am until 2:30pm the ladies get together to work on a variety of projects, including quilts, tie comforters, cards and of course, to visit! All of the projects are donated to fundraising events in the community. One of their beautiful quilts is always a hit at the Patchwork Benefit and Geauga Benefit Auction every year. Bring a lunch with you and join us soon.

We are very grateful to Anna Miller who joined DDC Clinic as our liaison within the Amish community, assisting us with our planned family meetings. She was able to begin immediately with planning the Mom's Coffee Break on Sept. 13th and the Family Fun Night on Sept. 22nd. Her help with these programs has been invaluable. **MOM'S COFFEE BREAK** and **FEELINGS OF MY HEART** mother's groups are meeting on the 2nd Wednesday of each month from 10:00am to 12:00pm. This is an opportunity for mothers of special needs children to get together to visit, share ideas and have a little time to relax! Coffee is provided and ladies are asked to bring a light snack. Babysitting is not available, but you are welcome to bring your preschooler with you! For more information call DDC Clinic at 440-632-1668.

Upcoming Dates for Mom's Groups Events:

Dec 13, 2017	Mom's Christmas Break and gift exchange
Jan 10	Feeling of My Heart and brunch
Feb 7	Mystery Trip
Feb 14	Mom's Coffee Break
Mar 14	Mom's Coffee Break
April 11	Mom's Coffee Break
May 2	Holmes County trip
May 9	Mom's Coffee Break

For more information call DDC Clinic at 440-632-1668.

DDC Welcomed Dr. Joseph Kingston

Dr. Kingston began his second year of residency at Aultman Hospital in Canton, Ohio.

He came to DDC Clinic in August for a 30 day rotation to learn about rare genetic disorders and to gain a better understanding of a Medical Home. Dr. Kingston is originally from Utah and lived in Texas prior to coming to Ohio. He is married with 5 children and hopes that his family will be joining him very soon in Ohio. He enjoyed his time at DDC and learning about the Amish culture. He even made time to join in the fun at two local auctions. We wish him well as he continues his education.

Immunization Program

The Vaccines for Children (VFC) Program helps ensure that all children have a better chance of getting their recommended vaccinations on schedule. Vaccines available through the VFC Program are those recommended by the Advisory Committee on Immunization Practices (ACIP). These vaccines protect babies, young children and adolescents from 16 diseases.

DDC Clinic began offering immunizations to the local community in 2009. The program continues to be a success and we are seeing our numbers increase every year. The number of newborns beginning the immunization process is especially encouraging. Through September 2017 we have provided 435 children with 1,067 vaccinations and 64 of them were receiving vaccinations for the first time.

Immunizations are by appointment only. Please call DDC Clinic at 440-632-1668 to schedule an appointment for your child. Appointments are made between the hours of 9:15am and 4:00pm.

2018 DATES WITH OPEN APPOINTMENTS ARE:

February 14th and 28th
March 14th and 28th
April 11th and 25th
May 9th and 23rd
June 13th and 27th
July 11th and 25th
August 8th and 22nd
September 12th and 26th
October 10th and 24th
November 14th
December 12th

Recent Publications

Patient-oriented research advances diagnoses and ground-breaking treatments, bringing relief to children and hope to their families. Sharing what we learn in serving children with special needs benefits all the world's children.

Clinical Rheumatology:
Tocilizumab reverses cerebral vasculopathy in a patient with homozygous SAMHD1 *mutation*

To view the full manuscript go to www.ddcclinic.org.

A child with special needs will inspire you to be a special kind of person.

Inherited, or Not Inherited?

Christine Wensel, MS, LCGC,
DDC Clinic Genetic Counselor

Inheritance of genetic traits and diseases is usually straightforward, or at least it seems that it should be this simple. Our genes code for all of our traits; how we look, how our body functions and whether or not we have a genetic disorder or disease. Our genes come in pairs. We receive half of our genetic make-up from our mother and half from our father. This sounds easy enough; but of course there are exceptions to this rule. This article will briefly cover a few basics of genetic inheritance and an exception to the rule.

Most of us are familiar with the idea that, for some disorders, a child with a genetic disease has inherited it from an affected parent. We have said or heard comments about a child inheriting a trait in appearance or health from one parent. Diseases that usually affect males and females equally and are inherited from an affected parent are called autosomal dominant disorders. A parent with an autosomal dominant disorder has one normal copy of the gene and one copy with a disease-causing mutation or variant. An affected parent may pass on either the normal variant of the gene or the disease-causing variant. Thus with each pregnancy, the chance that an affected parent will have an affected child is 50% or 1 in 2, like flipping a coin.

There are many genetic disorders for which an affected child does not have an affected parent; instead, the healthy parents are each carriers of the disease. Such genetic diseases are called autosomal recessive. Children with an autosomal recessive disorder have two copies of disease-causing variants in the same gene. The parents are healthy because even though they carry one disease-causing variant, they each have one normal variant as well. The single normal variant in this case is all that is needed to ensure the parent is healthy. Carrier parents may have a family history of the disease and worry that they could be a carrier but they rarely know for certain unless they have an affected child, multiple affected children or have had carrier testing. For autosomal recessive disorders, these carrier parents have a 25% or 1 in 4 chance that with each pregnancy both parents will pass the disease-causing variant to an affected child.

Genetics can be tricky when, all of a sudden, healthy parents have a child with a disorder known to be an autosomal dominant disorder, the type which is usually passed on from an affected parent. We know our basic genetics and this does not follow the rules. Remember the exceptions that were mentioned? One such exception to the rules of genetic inheritance is the *de novo* variant. *De novo* refers to a new change in the genetic material.

De novo variants happen all the time. Recent studies suggest that such changes occur many times in every single person, either prior to conception in the egg or sperm or very early in development before birth (and even through adulthood but that is a whole other story for another day). Most of these *de novo* changes go unnoticed and undetected as they cause no health or medical problems. However, if that new change occurs in a disease-causing gene it can lead to serious consequences in a child.

Part of the confusion caused by a *de novo* variant is that typically when the parents are healthy and the child is affected, we may automatically assume that the disorder is autosomal recessive. After all, this fits the inheritance pattern seen in autosomal recessive disorders. An additional layer of confusion occurs when multiple children are affected with this *"de novo"* disorder. *De novo* refers to a new genetic change; how can this be new when there are multiple children affected?

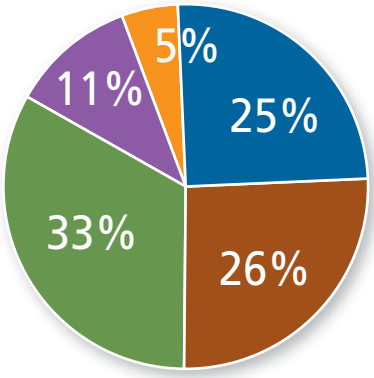
De novo changes actually may be found in a parent, but in only some cells of the body, not all. In such a case the parent may not show any sign or symptom of the genetic change. If the change is found in sperm cells or eggs then this *de novo* change can then be passed on to one child or to multiple children. Thus, an unaffected parent can have children who clearly show signs of an autosomal dominant condition.

By performing genetic testing combined with research testing as needed, DDC Clinic Molecular Diagnostics Laboratory has proven *de novo* variants to be the cause of a genetic disorder within several families. For some families, the *de novo* variant was inherited from an unaffected parent who carried the variant and passed it on. For other families, the *de novo* variant was found in just one affected child and not inherited from a parent. Once this *de novo* variant is part of the genetic make-up of an individual, it can then be passed on to future generations following the basic rules of genetics.

2016 Annual Report

Revenue

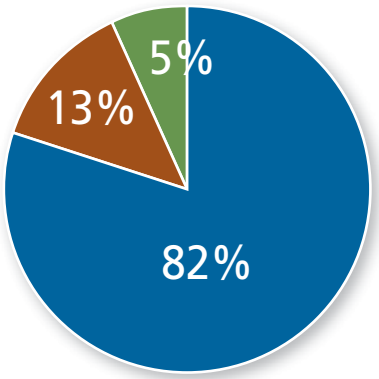
Total Revenue for 2016 – $1,244,374



Grants and Research	$	309,956
Benefit Auctions	$	329,741
Clinical Services	$	404,729
Individual Giving	$	139,468
Miscellaneous Income	$	60,480

Expenses*

Total Expenses for 2016 – $1,277,914



Program Services		$1,041,688
Fundraising	$	166,128
Management & General	$	70,098

**Reported less $97,089 depreciation.*

Statement of Financial Position

Assets		2016
Cash and Cash Equivalents	$	$540,256
Accounts Receivable	$	$140,698
Pledges Receivable	$	$850
Grants Receivable	$	$15,002
Prepaid Insurance	$	$25,732
Property and Equipment	$	$1,712,674
Cash restricted to Endowment	$	$392,393
Pledges Receivable	$	$250,000
Total Assets		**$ 3,077,605**

Liabilities and Net Assets

Accounts payable	$	2,425
Total liabilities	$	2,425
Net Assets		
Undesignated	$	2,279,484
Board designated	$	108,443
Total unrestricted	$	2,387,927
Temporarily restricted	$	153,303
Permanently restricted	$	533,950
Total net assets	$	3,075,180
Total Liabilities and Net Assets		**$ 3,077,605**

Thank you to our donors

Donor Recognition report from 1/1/2016 – 12/31/2016

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($25,000 and up)

The Char and Chuck Fowler Family Foundation
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Shiloh Mennonite Community Benefit Auction
Spartansburg Community Benefit Auction
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($10,000 - $24,900)

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It is only with your partnership and support that we are able to continue to do this most privileged work!

Thank you for your partnership!

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Center for Special Needs Children

14567 Madison Road
Middlefield, Ohio 44062

440-632-1668
ddcclinic.org

Patchwork
2018 BENEFIT

An evening to celebrate DDC Clinic for Special Needs Children

Save the Date!

APRIL 14, 2018

Join us at
The Federated Church Family Life Center
16349 Chillicothe Road
Chagrin Falls (Bainbridge Township), OH 44023

Enhancing the quality of life for people with special needs caused by rare genetic disorders.

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Help Our Special Children

Purchase a 2018 DDC Clinic Calendar and help special needs children in our community.

Call 440-632-1668 to purchase a calendar

Your $100 purchase enables DDC Clinic to provide much-needed medical care to children affected by rare genetic disorders.

With your help, we can make a difference in the lives of these special children and their families.

Thank you for your support.

Benefit Raffle Ticket Included

Each calendar includes a ticket to DDC Clinic's year-long benefit raffle.

Monthly winners may choose from the following prizes, each valued at $500 - $1,000.

- Pony cart
- Poly Vinyl 2-person glider
- Bernina sewing machine
- CL Clipper push mower
- Husqvarna 36-volt battery-operated weed trimmer and chainsaw
- Weber propane grill
- Rotating clothes line
- Ladies gift basket – gift certificates for fabric stores, groceries and stamping
- Maytag Wringer Washer
- DeWalt 20-volt 5-tool combo kit