Brandeis University

The Development of a Family History Collection Tool for use in a Pediatric Practice

A Pilot Study

Master's Thesis

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By
Cassandra Leduc

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Abstract

The Development of a Family History Collection Tool for use in a Pediatric Practice

A thesis presented to the Biology Department
and Genetic Counseling Program

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Cassandra Leduc

A complete family medical history is a valuable tool for understanding a person's genetic risks but it is often underutilized in the primary care setting. Methods for effectively obtaining and interpreting this information are not well-established. As obtaining a genetic family history may become standard practice in routine health care settings, it is necessary to have methods to effectively and accurately acquire the desired genetic family history. The purpose of this study was to design and pilot a pediatric family history collection tool in the form of a symptom-based checklist, to be used in a primary care setting. This checklist was piloted in a suburban pediatric practice located outside of Boston, MA for a two-week period. Parents who completed the checklist and practitioners who reviewed the family history information provided by the parents were invited to participate in a brief survey to assess the effectiveness of the tool.

A total of 159 families and 8 practitioners from the pediatric practice completed the survey. Over 97% of parents reported a positive impression of the checklist. Parents felt it was effective at collecting family history information and over 90% felt the amount of time it took them to complete the checklist was appropriate. Practitioners were more

divided in their thoughts about the checklist's success. Half viewed the checklist as an improvement over tools used previously by the practice, and most felt it collected relevant family history information. However, practitioners were concerned about the time and level of effort it required of them. It is unclear whether this concern would extend to any family history tool. Incorporating the checklist information into the patient's electronic medical record and educating parents and practitioners about the value and purpose of collecting a family history may improve overall satisfaction.

Keywords: family history, pediatrics, primary care, family history collection tool, genetics

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Introduction

The completion of the Human Genome project has brought with it increasing knowledge about the genetics of disease. With this growing information, it has become important for physicians in general practice to consider the significance of family medical history in providing the best preventative care and management of their patients. While taking a complete family medical history is a valuable tool for understanding a person's genetic risks, it is often underutilized in the primary care setting and methods for effectively obtaining and interpreting this information are not universally well-established. Historically, clinical genetics has been a specialty field in medicine dealing with a set of relatively rare disorders. Now, genetic knowledge is expanding beyond single gene disorders to an understanding of the genetic contributions to common complex diseases. Thus, physicians are beginning to move away from only monitoring people with a genetic disease to treating individuals with inherited risks for disease (Cheng et al. 2008). As a result, the limited number of genetic specialists cannot address the growing number of issues that come with this increased knowledge and genetics is extending into mainstream health care. Given this shift, the incorporation of a genetic family history component into the primary care setting takes on an even greater significance.

Trotter and Martin (2007) discuss the importance of incorporating family history into primary care, describing it as a "tool for public health and preventative medicine." A complete, updated three-generation family history could provide information to reduce risks, prevent concerning medical conditions, and identify genetic disease early for a

particular patient (Trotter & Martin, 2007). In addition, the medical family history is a useful tool for identifying at-risk family members, determining inheritance patterns and risks, and aiding in the diagnosis of a particular syndrome or condition (Beery & Shooner, 2004). Family history is known to be an important part of a patient's medical history and is a key element that physicians must account for when billing, but this information needs to become more complete and standardized in practice (Trotter & Martin, 2007).

Family history information is typically obtained through a discussion between the health care professional and patient during the visit, or through a brief questionnaire that the patient fills out prior to the visit (Qureshi et al., 2005; Trotter & Martin, 2007). A good family history tool must be quick, straightforward, easy-to-use and flexible enough to be used in multiple different settings and situations (Beery & Shooner, 2004). Quershi and others (2005) developed a family history questionnaire that could be used in situations where the time to take a full genetic pedigree is not available. Their questionnaire allowed a systematic way of obtaining family history data that could be used by health care providers who were not specialists in genetics (Quershi et al., 2005). To test the validity of using a family history questionnaire, Quershi and colleagues (2005) set up a comparative study with a group of patients that initially filled out the family history questionnaire and later participated in a genetic pedigree interview. They found that there was a 77 percent agreement between the genetic risk information obtained from both methods and that both instruments were equally effective in determining ethnicity-based risks of common recessive disorders (Quershi et al., 2005). Consequently, they

report that particularly in a primary care setting, the family health questionnaire can be an effective tool for collecting useful family history data in a systematic manner.

Pediatricians have a unique role in obtaining the medical family history for several reasons. They typically see their patients frequently in early childhood for check-ups and have a theoretical opportunity to obtain and update the family history during these "well child" visits (Trotter & Martin, 2007). Trotter and Martin (2007) recommended that a pediatrician take an extensive family history at the initial new patient visit and then update it at each visit in the future. Additionally, pediatricians have the benefit of being able to follow their patients from birth through adulthood, which allows the ability to intercede in the progression of some typical adult onset multi-factorial conditions, such as heart disease and certain cancers, before they become an issue (Cheng et al., 2008). Collecting a family history also has the potential to identify other childhood-onset disorders in the family, which may have implications for management and educational planning for that family. Pediatricians have already been exposed to newborn screening for their patients, and including a genetic family history to look for future concerns seems to be a relevant addition to their standard of care (Rosas-Blum et al., 2007).

There are several primary reasons that genetic family histories are not properly obtained: time, genetics knowledge, communication and awareness (Beery & Shooner, 2004; Rosas-Blum et al., 2007, Trotter & Martin, 2007). In the clinical setting, there is limited time available to meet with patients, so a family history must be obtained and reviewed in a short period of time (Trotter & Martin, 2007). Studies show that seventy percent of physicians feel that they don't have enough time to inquire about a patient's

family history during a visit and the average amount of time that a family physician spends on these discussions is less than three minutes in length (Acheson et al., 2000; Trotter & Martin 2007). Additionally, some primary care physicians do not see the relevance of obtaining a family history within their practices. In a small study by Trinidad and colleagues (2008), two-thirds of physicians did not feel that genetics was highly relevant to their everyday practice, only factoring in its importance for the cases where a patient has a known genetic condition. Some physicians also report that because genetic concepts are more difficult to explain, they aren't comfortable enough with their knowledge of genetic risks and are therefore less likely to include them in practice (Rosas-Blum et al., 2007). A majority of physicians (58%) mentioned that their own uncertainty and discomfort with the genetic information is a barrier to including genetic information in their daily practice (Trinidad et al. 2008).

As more is known about genetics, and the implications of this information have become better understood, the role of the genetic family history has developed into a critical piece of information for an individual's health management. Given that obtaining a genetic family history may well become standard of care for routine health care settings it seems worthwhile to develop the tools necessary to get the desired genetic family history in a simple yet productive manner. Existing family history collection tools are too broad, too lengthy, and not specific for use in a pediatric practice. Our study designed a focused pediatric family history collection tool to help obtain a comprehensive family history in a systematic manner and evaluated its success from both the patient and practitioner standpoints.

Methodology

Design of Family History Collection Tool

To determine the most logical design for our family history collection tool, we reviewed several existing family history questionnaires available online as well as the existing family history form available from the Westwood-Mansfield Pediatric Associates (

Table ***1***). We specifically evaluated these forms on their length, apparent ease of use, focus and content.

Table 1: List of family history collection tools evaluated to determine the best design and content for our pediatric family history checklist

Family History Collection Tool (Creator)	**Obtained from:**
My Family Health Portrait (US Surgeon General)	http://www.hhs.gov/familyhistory/
Preconception/ Prenatal Family History Questionnaire (March of Dimes)	http://www.marchofdimes.com/files/GYP_PrenatalQuestionnaire.pdf
Family History (Westwood-Mansfield Pediatric Associates)	Westwood-Mansfield Pediatric Associates office

After evaluating several existing family history forms that were deemed too lengthy, too vague, or too complicated, we aimed to develop a new family history collection tool for use in a pediatric practice. The resulting tool (Appendix A) was designed as a checklist that can be easily filled out by the parent(s)/guardian(s) of pediatric patients in advance of their appointment. The family history checklist consists of a list of various genetic conditions and features with check boxes and space to indicate

the affected family member. We were careful to design a tool that would be easy-to-use and succinct, while still being able to gather comprehensive genetic family history information for the patient. Additionally, since we were working with a pediatric practice, we focused our family history collection tool on genetic conditions that would likely present during childhood.

Instead of focusing specifically on genetic disorders, we designed the family history collection tool around specific symptoms or features of certain conditions to make it more patient-friendly. In addition, the design of the tool took into account concerns from the pediatricians regarding a level of discomfort with, and limited knowledge of, genetic disorders. To improve practitioner satisfaction and help them interpret the family history checklist for their patients, we created a follow-up guide for use in connecting the family history data back to a suspected genetic disorder (Appendix D). While this follow-up guide cannot cover every genetic syndrome with a given feature, it was developed to suggest the most likely possibilities based on certain checked features in a family history.

Pilot Period

The Brandeis University IRB granted us approval through expedited review for a period of one year effective starting January 23, 2009. The family history checklist was piloted in the Westwood-Mansfield Pediatric Associates offices for two weeks, from February 26, 2009 to March 11, 2009. The group is a suburban private pediatric practice in Metrowest Boston with seven pediatricians and eight nurse practitioners and a patient population of 11,000-12,000. We piloted the new family history checklist with the parents whose child had either their initial visit or standard well-child visit during the two week pilot period.

The practice staff mailed the family history checklist to all those patients in advance of their upcoming appointments. At the end of the checklist was a feedback survey for parents to comment on various aspects of the checklist. Parents were called and reminded about their upcoming appointment and notified that the family history checklist and feedback survey were also available through the practice website (http://wmpeds.com/forms/familyhistory.pdf). The practice staff instructed parents to fill out the family history checklist and bring it with them to their child's appointment. The medical receptionists collected the family history checklist and survey when the patients were checked in. The feedback survey was separated from the family history and set aside. The family history checklist was put into the patient's chart for the pediatricians or nurse practitioners to review to enable them the opportunity to discuss any concerns with the patient and/or family.

Data Collection

Feedback was collected from both the parents who completed the new family history checklist as well as from the nurse practitioners and pediatricians of Westwood-Mansfield Pediatric Associates. We surveyed both groups, each with a different survey, to gather information about the family history checklist's practicality, strengths, and weaknesses.

The brief parent feedback survey at the end of the family history checklist (Appendix B) consisted of 7 Likert scale and 3 open-ended questions about their experience with and opinion of the family history checklist. All parents with appointments between February 26, 2009 and March 11, 2009 were given the opportunity to complete the feedback survey. The parent feedback survey was separated from the

collected family history data by the office staff and we did not have access to the patient's family history data. The parent feedback data responses were coded and inserted into Microsoft Excel for data analysis.

Towards the end of the pilot period, the practice coordinator e-mailed the pediatricians and nurse practitioners to invite them to participate in an online feedback survey which was hosted at surveymonkey.com. This survey utilized 10 Likert scale and 4 open-ended questions to gather practitioner input about issues such as the practicality of the family history questionnaire, ease of use, length appropriateness, helpfulness of the information collected, things they would change and overall thoughts about the family history collection tool in general. The survey remained open on surveymonkey.com from March 9, 2009 to March 19, 2009 to allow practitioners to respond at their convenience. We sent out reminder e-mails to all providers on March 11, 2009 and March 17, 2009 with a link to the survey to encourage the practitioners to participate in the online survey before it was closed. We used surveymonkey.com for the preliminary analysis and exported the collected data into Microsoft Excel for additional data analysis.

Feedback data from both the parents and practitioners were analyzed manually to look for trends and correlations. We determined the percentage of participants who selected each response, and used these values to evaluate the overall success of the family history collection tool from the provider and parent perspective.

Results

Through our evaluation of existing family history collection tools, we identified several strengths and weaknesses for each tool. A summary of the pros and cons for the three evaluated tools is listed in Table 2.

Table 2: Evaluation of Existing Family History Tools

Tool	***Pros***	***Cons***
My Family Health Portrait	• collects data from patient and prints family history summary • breaks more common conditions into categories, allows patients to specify additional disorders in family members	• adult focused • may be too general to pick up rare genetic conditions • doesn't ask about extended family (3rd degree relatives) unless patient specifically chooses • requires patient to select each family member to add a disorder or condition • printable version does not list any conditions – just requires patient to list conditions for each relative
March of Dimes Family History Questionnaire	• checklist format for ease • asks patient to specify relationship and affected side of the family • good mixed list with common disorder features and specific syndromes	• prenatal/preconception focused • may be too time consuming and lengthy (3 pages long)
Westwood-Mansfield Family History Form	• somewhat pediatric focused • brief family history (2 pages), probably not too time consuming patient	• may not have enough guidance to pick up rare genetic conditions • first page with list of conditions does not ask for specific family member • second page requires patient to determine which conditions are important to list

Parent Feedback

A total of 536 families had access to the family history checklist during the pilot period. Of those parents (number) that filled out the family history checklist, 166 went on to fill out the parent feedback survey giving us a response rate of 31%. Several respondents only answered one or two survey questions and skipped the rest of the survey. To help balance our data pool, we eliminated all survey respondents who did not answer at least three of the seven Likert-scale survey questions. This gave us a final respondent tally of 159, for a response rate of 30%.

We asked parents to rate their overall opinion of the family history collection tool. As shown in Figure 1, greater than 97% of respondents had a favorable impression of the checklist with 150 out of 154 indicating that they felt that the family history checklist was either okay or that they liked it.

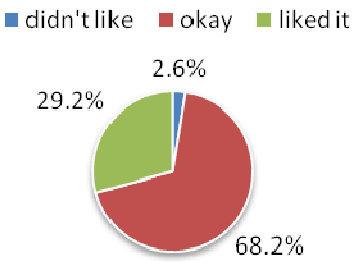

Figure 1: Parents' overall impression of the family history checklist

Of the parent respondents that liked the checklist and went on to comment about its most valuable aspect, several indicated that the most valuable aspect was the ease in filling out the checklist. One of the parents, who responded that they didn't like the family history checklist, stated that the checklist was "hard to fill out as if I were child" and went on to

explain that the part where they had to indicate the relationship to the child was "unclear despite directions."

We also asked parents to indicate their opinion of the success of the checklist at collecting family history information (Figure 2).

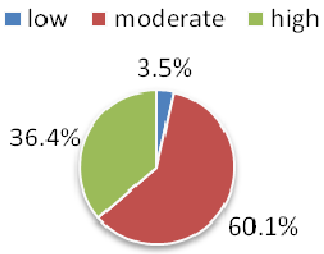

Figure 2: Parents' opinion of the success of the checklist at collecting family history

Most parents, 86 of 143 respondents (60.1%), felt that the family history checklist was moderately successful at collecting this information, and a significant number, 52 of 143 (36.4%) thought that the checklist had a high level of success. Only 3.5% of parent respondents indicated that they felt the checklist had a low success rate for collecting family history information, but none of these respondents went on to comment further when asked about the problems with the form or desired changes. Of the parents that felt the checklist was moderately successful, there were mixed reviews about the checklist's completeness. Several respondents commented that the checklist was "possibly missing information" with one respondent stating that they "were not sure about the completeness." Other respondents commented that "the checklist showed common problems on both sides of family" and that it "covered many items."

As shown in Figure 3, 23% of parent respondents (31 of 135) felt that the family history checklist was missing information while 77% felt that the checklist was very complete. Parents commented that the checklist was "missing cancer over 40 years," "missing other heart problems" and did not include a "place to list patient's information." Additionally, parent respondents commented that the form was missing high blood pressure and cholesterol problems as well as alcoholism.

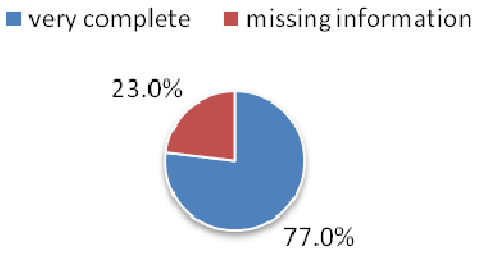

Figure 3: Parents' opinion on the completeness of the checklist at collecting family history data.

We also asked respondents to comment on the amount of time they spent filling out the family history checklist and their ease with filling it out. Over 90% of parents, 140 of 155 respondents, felt that the checklist took just the right amount of time to fill out. Four respondents (2.6%) felt that not enough time was spent filling out the family history checklist and 11 (7.1%) felt that it took too much time to complete. Just under 95% of the parent respondents, 148 of 157, felt that the family history checklist was just the right level of difficulty with only one parent feeling that the checklist was too simple.

Parents also rated their personal knowledge of their family history, with the majority (95 of 157) of parents classifying themselves as having an average knowledge, and 53 of 157 parents reporting a very high knowledge of their family history (Figure 4).

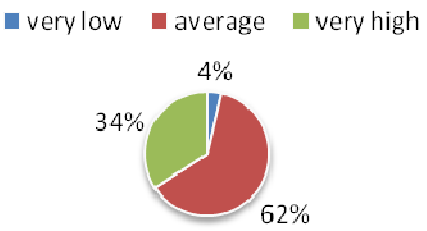

Figure 4: Parents' reported knowledge of their family history.

Among those parents who reported having a very low knowledge of their family history, no one reported problems with filling out the checklist and several individuals commented that there was nothing they would change. One parent commented on her limited family history knowledge in relation to the family history checklist, saying "in general [I] found the info requested to be relevant and need to do better job of finding accurate answers."

The survey asked parents to provide feedback about the number of family history questions that were hard to understand. The vast majority, 133 of 140 respondents (95%) indicated that less than 5 checklist questions were unclear. No parents responded that there were 15 or more questions that were hard to understand, and 7 of 140 (5%) felt that between 6 and 14 family history checklist questions were unclear.

We also asked parents to comment on the aspects of the family history checklist that were most valuable to them. One parent commented that the checklist "increases your own awareness of risks" and another parent similarly commented that they "like knowing that full family history is part of [their] medical file." An additional parent remarked that they "did not find this [checklist] to be of any value to me." Respondents

also stated that the checklist "covered many items," "was quick to use" and was valuable because it "included developmental as well as physical and genetic" information.

When asked about things they would change about the checklist, or problems that they had with the family history checklist, many parents commented that they had none. One parent commented that it was "too general" and needed "more specifics." Some parents commented about changes to the structure of the family history checklist, with one respondent suggesting that we "alphabetize the checklist" and another suggesting that we provide "more space to write specifics." One mom commented about the need to consider alternative families, stating that her family "has two moms and our daughter was conceived by donor." Additionally parents commented that they were unsure about how this family history information was useful or valuable for their child's health.

Practitioner Feedback

All nurse practitioners and pediatricians of the Westwood-Mansfield Pediatric Associates were invited to participate in an online feedback survey. Of the fourteen practitioners, we received 10 responses to our online survey providing us with an overall response rate of 71%. However, two of these respondents did not provide feedback beyond the initial demographic questions, which left us with eight respondents who completed the feedback survey, three nurse practitioners and five pediatricians. We first asked the practitioners to describe their knowledge of pediatric genetic conditions and all of the nurse practitioners and pediatricians (8 of 8) rated their knowledge as "average".

We also asked respondents to provide their overall impression of the family history collection tool. Of the 7 respondents what chose to answer, 3 respondents (42.9%) thought the tool needs improvement, 2 respondents (28.6%) rated the tool as mediocre

and the remaining two respondents (28.6%) thought the tool exceeds expectations. To determine if nurse practitioners and pediatricians felt differently about the new family history collection tool, we cross tabulated the results in survey monkey, looking at the responses of pediatricians and nurse practitioners separately (Table 3).

Table 3: Overall impression of this family history collection tool based on the respondent's identified role in the practice as either a nurse practitioner or pediatrician.

Response	**Nurse Practitioners**	**Pediatricians**
needs improvement	0 (0.0%)	3 (75.0%)
mediocre	1 (33.3%)	1 (25.0%)
exceeds expectations	2 (66.7%)	0 (0.0%)
total respondents	3	4

Of note is that while 2 out of 3 responding nurse practitioners felt that the new family history collection tool exceeds expectations, 3 out of 4 pediatricians who responded felt that the tool needs improvement.

To get a better sense of the success of the family history checklist in practice, we asked the pediatricians and nurse practitioners to compare it to their previous family history form. When asked about their opinion of the family history checklist, half of the respondents (4 of 8) felt that it was better than the practice's previous family history forms (Figure 5). The remaining practitioners did not see the new checklist as an improvement with 25% feeling that it was about the same as the previous form and the other 25% feeling that the new family history tool was worse than the previous one.

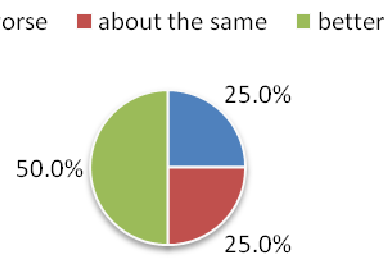

Figure 5: Practitioners' opinion of the new family history checklist compared with the practice's previous family history forms

Practitioners also had the opportunity to comment on both the success of the tool at collecting family history and the usefulness of information collected to patient care and health management. Seven out of eight respondents (87.5%) classified the tool as having moderate or high success at collecting family history information. Additionally, seven of eight respondents (87.5%) rated the usefulness of the collected information as somewhat or very useful for patient care and health management. Only one pediatrician indicated that the tool had both a low level of success at collecting family history information and also that the information collected was not very useful. This pediatrician went on to comment on the family history checklist, stating that parents "fill it out wrong at least as often as they get it right, and the information recovered [from parents] is too vague."

The survey also asked about how often the practitioners used the family history collection tool to discuss concerns at the time of the patient's appointment. One pediatrician commented that they frequently used the tool to discuss these concerns. Of the remaining seven practitioners, three respondents indicated that they did not use the checklist to discuss concerns very often, and four indicated that they sometimes used the family history checklist to discuss concerns at the appointment. We also asked the practitioners about the usefulness of the family history follow-up guide. Half reported

that they did not use the follow-up guide. One respondent indicated that the guide was very useful and three respondents felt that the guide was somewhat useful in practice.

When the providers were asked about their perceived ease of incorporating the family history collection tool into their practice, the majority of respondents (5 of 8) felt that it would require a moderate effort (Figure 6). Two practitioners felt that the family history checklist would be easy to incorporate and the remaining practitioner felt that it would be difficult to incorporate into practice.

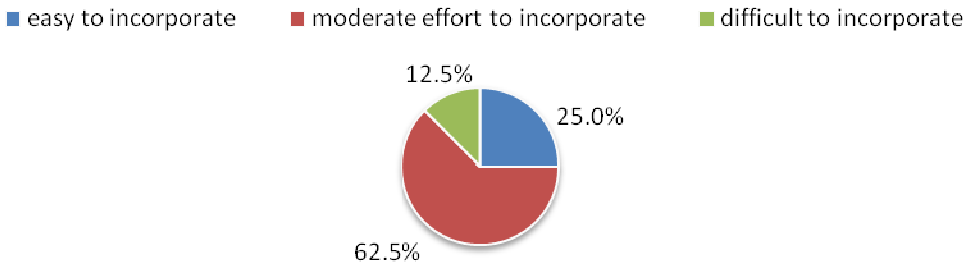

Figure 6: Practitioners' perceived ease of incorporating family history checklist into practice.

Practitioners were more split when asked to indicate their desire to continue using the family history checklist (Figure 7). Three practitioners indicated a high desire, three indicated a medium desire, and two indicated a low desire to continue using the checklist.

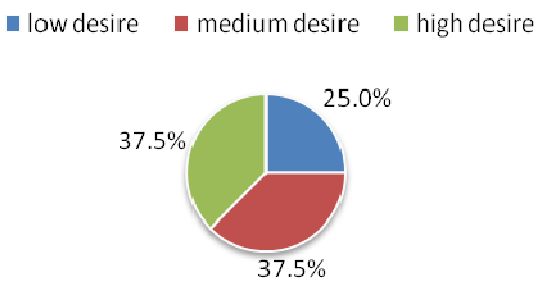

Figure 7: Practitioners' self-reported desire to continue using the family history tool.

Among the nurse practitioners, two of the three respondents expressed a high desire to continue using the family history checklist and the other respondent expressed a medium desire. Among the pediatricians however, only one of five respondents indicated a high desire to continue using the family history checklist with other four respondents split evenly between a low and medium desire.

We lastly asked the practitioners to comment on the aspects of the family history checklist that they found most valuable as well as most problematic. One nurse practitioner commented on the value of the new family history checklist, saying it "was a more pointed, specific tool than our previous [family history] form." That nurse practitioner went on to comment that it "allows us to focus in on aspects of history that are relevant instead of parents trying to remember what medical conditions might have been in the family." Another nurse practitioner commented that the family history checklist was "very user friendly" and went on to comment that the most valuable aspect was "the part where the parent had to circle the correct side of the family and then write in the affected family member."

Some practitioners also commented on aspects of the tool that were not so successful. Several respondents felt that the design of the checklist was not ideal. One pediatrician commented that the family history checklist was "difficult to read [because]

lots of info crammed together" while another commented that "it is very dense and cumbersome." Additionally, one nurse practitioner stated that "the tools appears at first glance as if it will be a lot of work for families" and recommended trying to "condense [it] to one page." One pediatrician also expressed dissatisfaction in the time required to review the information and input it manually into the patient's medical record. This pediatrician commented that "if it were automatically incorporated into the record, that would be better" and stated that the other practice family history form "was automatically input by his office staff."

Discussion

Our evaluation of existing tools highlighted the need for a simple, focused, family history collection tool for use in the general pediatric practice setting. It revealed that existing tools were not ideal for use in a pediatric practice and identified valuable components which we used to develop our family history collection tool. We took aspects of these existing family history forms and combined them to create our resulting family history checklist, which we feel is better suited for the pediatric practice setting. We created our family history checklist to be completed by the patients' parents in advance of their appointment and purposely kept the checklist broad to collect the most information about the family history. When we presented the office with our family history tool, there was concern that the practitioners would not feel comfortable interpreting the parents' checklist responses, so we additionally developed a follow-up guide to correspond with our family history checklist to improve practitioner satisfaction.

A total of 536 patients were seen and had access to our family history checklist during our two week pilot period. Some of these parents had recently filled out the Westwood-Mansfield Pediatrics family history form and chose not to complete our family history checklist. Therefore, the actual number of parents with whom we piloted our family history checklist is somewhat lower. Of the parents that completed our family history checklist, 166 parents also provided feedback about the family history checklist for a response rate of over thirty percent.

During our pilot period, there was some confusion among the office staff about the swapping over of the family history tools. Consequently, at least some patients

received both the practice's existing family history forms as well as our newly developed family history checklist. We are unsure of the exact number of patients affected by this situation, but it is worth briefly considering the potential impact this may have had on their responses to our survey when asked to evaluate the family history checklist. For the purposes of this discussion, we assumed that the vast majority of respondents were evaluating our piloted family history checklist and used the survey responses to guide our evaluation of the checklist.

Overall, the survey data from the families showed that our family history checklist was well received by the parent respondents. Less than three percent of parent respondents had a negative impression of the checklist and indicated that they didn't like it with nearly seventy percent of parents rating the checklist positively. Additionally, most parents felt that the tool was at least moderately successful at collecting family history and that it was the right length and difficulty level. These results show that our newly designed checklist is able to gather family history information while meeting the needs for a simple to fill out and concise family history collection tool. While these results are promising for the future of the family history checklist, we cannot rule out some level of an ascertainment bias. It is possible that parents who felt the family history checklist was already too long may not have gone on to fill out the additional feedback survey page so our results may be skewed towards positive responses.

Additionally, we assessed the parents' opinion of the completeness of our family history checklist. Slightly less than one quarter of respondents felt that the family history checklist was missing information such as cancer over the age of 40, alcoholism, and hypertension. Most of the conditions that parents felt were missing were purposely left

out to tailor our family history checklist to risks for pediatric-onset conditions. Providing parents with more information about why we were collecting this family history information may be helpful in raising awareness and helping parents feel more comfortable with the completeness of the checklist. Additionally, several parents did not see the value of filling out the family history checklist for their child's health management and one parent felt that the checklist was not ideal because it did not consider alternative families. These problems can be addressed by more clearly communicating the purpose and benefits of collecting a biological family history in the assessment of potential genetic risks for the pediatric patients so that parents feel more connected to the collected family history information.

Our pilot study showed that practitioners are much more divided in their opinions of the family history checklist and provide mixed reviews. Practitioners as a whole were fairly divided between feeling that the tool needs improvement, rating it as mediocre, and feeling that it exceeds expectations. Some practitioners felt that our new family history checklist required too much time to sort through the patients' responses to determine which, if any, conditions to be concerned about, whereas some felt that our checklist was a better, more targeted mechanism for collecting family history data.

When we analyzed the opinions of nurse practitioners and pediatricians separately, the nurse practitioners appeared to have a much more positive opinion of the family history checklist than the pediatricians. The nurse practitioners who responded to our survey seem more satisfied with the family history checklist and therefore more likely to continue using it in their practice. Most pediatrician respondents stated dissatisfaction with the family history checklist and indicated that the tool still needs

improvement before it is practical to fully incorporate into their practice. However, while the apparent differences between the opinions of nurse practitioners and pediatricians are interesting, our analysis is limited by the size of our practitioner sample. Our small sample sizes of 3 nurse practitioners and 5 pediatricians from one private pediatric practice make it impractical to draw any significant conclusions about differences of opinion between pediatricians and nurse practitioners as a whole. Future studies should evaluate our family history collection tool in additional pediatric practices to broaden the scope of feedback.

Overall, the majority of practitioners felt that the family history checklist is successful at collecting family history information and that the information collected is of some value for the patient's health management. These responses indicate that our checklist was properly designed for its goal of collecting useful family history information and that perhaps addressing some stylistic problems would improve practitioner satisfaction with our checklist. Some concerns about the family history checklist were the amount of time the checklist requires for physicians to interpret it as well as its length and overall design. Some practitioners also worried that the family history checklist may be too cumbersome for their patients although our parent responses did not support this concern and indicated that the checklist was a good length and difficulty.

Interestingly, all eight practitioners rated their knowledge of genetic conditions as average so we were not able to compare differences in opinions of the family history checklist based on the practitioners' self-reported knowledge of genetics. We considered several explanations that may account for the lack of variation in responses and reasons

why the practitioners may have chosen the middle ground. One possibility is that the question is worded vaguely with only the option of having a very low, average, or very high knowledge level. Perhaps if we had asked about high or low, and eliminated the extreme end of very high or very low, we would have seen some different responses. Another possibility is that practitioners "do not know what they do not know" and may not realize the number of genetic conditions that they are unaware of until a situation arises where a given genetic condition, with which they are unfamiliar, presents itself.

Recommendations

Our pilot study showed that the parents are relatively satisfied with the family history checklist. Based on the parental responses, there does not appear to be a significant need to alter the design or content of our family history collection tool. However, in future versions of this tool, we may want to provide parents with a more detailed introduction to explain the reasoning for collecting family history of biological relatives and the focus on genetic conditions known to have a pediatric onset. This additional information may improve the parent satisfaction with the family history checklist and better educate them about the importance of family history in individual health management.

Although we had some success in the design of a family history checklist, the results of our provider surveys indicate the checklist needs modifications to meet the needs of practitioners. To improve practitioner satisfaction, the family history checklist must become less time consuming for practitioners. While not within the scope of this pilot project, the ideal family history collection tool would include a family history checklist component that could be electronically filled out by parent or inserted into

electronic medical record by office staff, as well as a computerized clinical support tool that would suggest genetic conditions, for which a patient may be at risk, based on the family history data (Rich et al., 2004). Future studies could work to develop a more automated approach to collecting, analyzing and storing family history information so that the process is less time consuming. In addition, a partnership between genetic professionals and computer programmers could design an automated program that could present the practitioners with a list of potential genetic conditions for which their patients may be at risk based on the input family history. Similar to the information collected for our practitioner family history follow-up guide, an automated family history analysis program could utilize information about genetic conditions from sites such as Online Mendelian Inheritance in Man (http://www.ncbi.nlm.nih.gov/omim) or GeneTests (www.genetests.org) to calculate a list of most likely conditions.

The practitioners in our study indicated that they have an average knowledge of pediatric genetic conditions. However, with the rapid expansion of genetics in medicine, there is always more to learn. One way to provide practitioners a means to further their genetic knowledge is by making available seminars geared toward their profession in which they can learn about and discuss potential genetic issues. We encourage genetic professionals and/or regional hospitals to consider having conferences focused on topics that may be encountered by pediatric primary care practitioners. Such conferences could serve as resources for these professional regarding the value of family history information and the various genetic conditions that may present during childhood.

Future studies should also reach out to more pediatric practices to gather information from a larger pool of practitioners and parents and have a more

representative population from which to draw conclusions about the success of the family history collection tool. It would be interesting to survey more nurse practitioners and pediatricians to see if their opinions of the family history collection tool are substantially similar to those determined from our provider surveys or if the provider opinions in this study are an artifact of our small sample size. In future studies, it may also be helpful to interview or survey nurse practitioners and pediatricians to assess both what type of seminar or conference they would be interested in attending and what other methods of enhancing their genetic education they would find useful.

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Appendix A: Family History Collection Tool

Patient's name: _____ DOB: _____
Parent's name: _____ Date: _____

To better understand your potential health risks, we ask that you please provide some information regarding your family history. **If you are the parent/guardian filling this checklist out, for the purpose of this family history, everywhere you see the word "you" or "your" please fill it out according to the relationship to your child**. Think about both sides of your family, including siblings, parents, grandparents, uncles/aunts, & cousins. Looking at the following list, please check off any conditions known to be present in the family and indicate their relationship **to your child**.

Please circle the correct side of the family (maternal/paternal) **and indicate the affected relative** (brother, mother, cousin, aunt, grandfather, half-sister, etc.)

developmental delay/mental retardation ☐ *maternal/paternal* _____
autism spectrum disorder (autism, PDD, Aspergers)☐ *maternal/paternal* _____
blindness or significant vision loss by age 40 ☐ *maternal/paternal* _____
cataracts/glaucoma by age 40☐ *maternal/paternal* _____
childhood diabetes ☐ *maternal/paternal* _____
congenital heart defect (ex. hole in heart) ☐ *maternal/paternal* _____
muscular dystrophy ☐ *maternal/paternal* _____
deafness or significant hearing loss by age 40 ☐ *maternal/paternal* _____
obesity ☐ *maternal/paternal* _____
depression ☐ *maternal/paternal* _____
mental illness (bipolar disorder, schizophrenia, etc.) ☐ *maternal/paternal* _____
cleft lip/palate ☐ *maternal/paternal* _____
tumors under the skin ☐ *maternal/paternal* _____
delayed/absent puberty ☐ *maternal/paternal* _____
neurological disorder ☐ *maternal/paternal* _____
cancer at a young age (<40) ☐ *maternal/paternal* _____
type: colon/breast/leukemia/other (please specify) _____ age at diagnosis _____
stillbirth/ neonatal childhood death ☐ *maternal/paternal* _____
individuals that died in infancy/childhood ☐ *maternal/paternal* _____
joint laxity/recurrent dislocations ☐ *maternal/paternal* _____
bleeding/clotting disorders ☐ *maternal/paternal* _____
genetic anemia/thalassemia ☐ *maternal/paternal* _____
unusual skin growths/conditions ☐ *maternal/paternal* _____
multiple or unusual birthmarks ☐ *maternal/paternal* _____
epilepsy/seizures ☐ *maternal/paternal* _____
energy problems/crises ☐ *maternal/paternal* _____
thyroid problems ☐ *maternal/paternal* _____

Patient's name: _____ DOB: _____

problems with muscle tone or muscle spasticity ☐ *maternal/paternal* _____
dental problems (absent/abnormally-shaped teeth) ☐ *maternal/paternal* _____
respiratory disease/chronic lung condition ☐ *maternal/paternal* _____
overgrowth disorder ☐ *maternal/paternal* _____
spinal problems (ex. scoliosis) ☐ *maternal/paternal* _____
abnormally developed hands/feet (ex: club foot) ☐ *maternal/paternal* _____
patches of different colored hair/skin ☐ *maternal/paternal* _____
iron overload syndrome (hemochromatosis) ☐ *maternal/paternal* _____
kidney problems (extra/missing/polycystic) ☐ *maternal/paternal* _____
multiple fractures/broken bones ☐ *maternal/paternal* _____
unusually tall/short for family ☐ *maternal/paternal* _____
smaller/larger than normal birth weight for family ☐ *maternal/paternal* _____

Is there anything in the family that you would like to discuss that we didn't already ask about?
_____

Please explain any checked conditions to provide additional information

Is it okay to discuss family history in today's appointment? yes☐ no☐
If no, how/when should this information be discussed? _____

---------- separate family history from feedback survey here ----------

Appendix B: Parent Feedback Survey

This family history checklist was designed by a graduate student at Brandeis University and is being piloted at the Westwood-Mansfield practice. The practice is collaborating with the graduate student to assess the effectiveness of the family history questionnaire. If you choose to participate in the survey below, the graduate student will receive your anonymous feedback (provided on this page) to evaluate the utility of the checklist but **will not have access to your family history information or medical records**. This feedback page will be detached from the family history data by the practice staff, so any opinions you provide will remain anonymous.

Participation in this survey is optional and provided feedback will be analyzed to identify the strengths and weaknesses of the family history checklist. This survey will not collect any identifying data from you and should take you less than 5 minutes to complete. You can skip any questions or end the survey at any time.

Thank you in advance for your time and the feedback you provide.

Please circle the answer that best describes your opinion of the following:

Your personal knowledge of your family history	very low / average / very high
Overall impression of the family history checklist	didn't like / okay / liked it
Success of the checklist at collecting family history	low success / moderate / high success
Time spent filling out the family history checklist	not enough / just right / too much
Ease of filling out the family history checklist	too simple / just right / too complicated
Number of family history questions that were unclear or hard to understand?	a few (<5) / some (6 – 14) / a lot (15+)
Completeness of checklist in gathering family history	very complete / missing information

What aspect(s) of the family history checklist did you find most valuable?

What aspect(s) of the family history checklist did you have problems with? Is there anything you would choose to change?

Any additional comments?

Appendix C: Practitioner Feedback Survey

Practitioner Feedback Survey *(constructed in survey monkey and a link to participate anonymously was e-mailed out to all nurse practitioners and pediatricians at the Westwood-Mansfield Associates)*

Opening page of survey:

Thank you for piloting this family history collection tool in your practice. Please take a few minutes to provide feedback about your experiences with the tool. The provided survey information will remain anonymous and identified personal history data will not be collected. The questionnaire should take less than 10 minutes of your time and will be helpful in evaluating the family history collection tool. The information you provide is useful in identifying the strengths and weaknesses of the family history collection tool in practice. Your feedback is optional and you will have the option to exit the survey at any time.

Thank you in advance for your time and any feedback you provide.

By clicking next below, I agree that I am a pediatrician or nurse practitioner with Westwood-Mansfield Pediatric Associates and wish to continue with the survey.

Page 2 of survey:

Please provide a little bit of information about yourself and your role with the Westwood-Mansfield Pediatric Associates.

Role in the practice:

__ nurse practitioner
__ pediatrician
__other (please specify)

_____

Years as a practicing pediatrician or nurse practitioner:

__ Less than 5 years
__ 5 to 9 years
__ 10 to 14 years
__ 15 to 19 years
__ 20 or more years

Page 3 of survey:

Please choose the answer that best describes your opinion of the following statements:

1. Your understanding of pediatric genetic conditions
   very low / average / very high

2. Overall impression of this tool
   needs improvement / mediocre / exceeds expectations

3. Success of the tool at collecting family history
   low success / moderate / high success

4. Ease of incorporating family history collection tool into your practice
   easy to incorporate / moderate effort to incorporate / difficult to incorporate

5. Percentage of patients (during pilot period) for which family history data was available to you at the time of the appointment
   very few / many / almost all

6. Usefulness of information collected to patient care and health management
   not very useful / somewhat useful / very useful

7. Comparison of new family history tool to previous family history tool
   better than previous tool / about the same / worse than previous tool

8. Usefulness of family history follow-up guide
   not very useful / somewhat useful / very useful / did not use

9. How often did you use the family history collection tool to discuss family history concerns at the time of the visit
   not very often / sometimes / frequently

10. Your desire to continue using family history collection tool
    low desire / medium desire / high desire

Final page of survey

Final Feedback comments

1. What aspect(s) of the family history collection tool did you find most valuable?

2. What aspect(s) of the family history collection tool did you have problems with?

3. Are there aspect(s) of the family history collection tool you would choose to change? Please describe.

4. Any additional comments?

Appendix D: Provider Family History Follow-up Guide

Provider Family-History Follow-up Guide – by Checklist Feature

This guide is not intended to be diagnostic, but provides some additional information about possible genetic syndromes based on the provided family history. For any given genetic syndrome, there are many features and this list only covers some of the more common symptoms/syndromes that might be picked up by family history. For additional information or if you have concerns about a patient's family history, please consult with a genetics professional.

- developmental delay/mental retardation
    - Tuberous sclerosis – autosomal dominant
    - Prader-Willi syndrome – depends on mechanism, usually sporadic
    - Angelman syndrome – depends on mechanism, usually sporadic
    - Noonan syndrome – autosomal dominant
    - Fragile X syndrome – x-linked (triplet repeat disorder)
    - Williams syndrome – autosomal dominant
    - Canavan disease – autosomal recessive
    - Smith-Lemli-Opitz syndrome – autosomal recessive
    - Wolf-Hirschhorn syndrome – generally sporadic
    - Zellweger syndrome spectrum – autosomal recessive
    - Sotos syndrome – autosomal dominant
    - Cornelia de Lange syndrome – autosomal dominant/x-linked
- autism spectrum disorders
    - Angelman syndrome – depends on mechanism, usually sporadic
    - Fragile X syndrome – x-linked (triplet repeat disorder)
    - Rett syndrome – x-linked dominant
    - Cornelia de Lange syndrome – autosomal dominant/x-linked
- blindness or significant vision loss by age 40
    - Retinitis pigmentosa – various modes of inheritance
    - Zellweger syndrome spectrum – autosomal recessive
    - mitochondrial disorders – various modes of inheritance
    - Bardet-Biedl syndrome – autosomal recessive
    - Usher syndrome – autosomal recessive
- cataracts/glaucoma by age 40
    - Marfan syndrome – autosomal dominant
    - Myotonic dystrophy – autosomal dominant
- childhood diabetes
    - mitochondrial disorders – various modes of inheritance

- congenital heart defect
    - Noonan syndrome – autosomal dominant
    - Williams syndrome – autosomal dominant
    - 22q11 deletion syndrome (VCFS/DiGeorge) – autosomal dominant
    - Wolf-Hirschhorn syndrome – generally sporadic
    - Sotos syndrome – autosomal dominant
    - Cornelia de Lange syndrome – autosomal dominant/x-linked
- muscular dystrophy
    - Duchenne/Becker muscular dystrophy – x-linked recessive
- deafness or significant hearing loss by age 40
    - Neurofibromatosis type 2 – autosomal dominant
    - Ectodermal dysplasia – various forms and modes of inheritance
    - Wolf-Hirschhorn syndrome – generally sporadic
    - Zellweger syndrome spectrum – autosomal recessive
    - mitochondrial disorders – various modes of inheritance
    - Cornelia de Lange syndrome – autosomal dominant/x-linked
    - Usher syndrome – autosomal recessive
- obesity
    - Prader-Willi syndrome – depends on mechanism, usually sporadic
    - Bardet-Biedl syndrome – autosomal recessive
- depression
- mental illness
    - 22q11 deletion syndrome (VCFS/DiGeorge) – autosomal dominant
- cleft lip/palate
    - 22q11 deletion syndrome (VCFS/DiGeorge) – autosomal dominant
    - Smith-Lemli-Opitz syndrome – autosomal recessive
- tumors under the skin
    - Tuberous sclerosis – autosomal dominant
    - Neurofibromatosis – autosomal dominant
- delayed/absent puberty
    - Androgen Insensitivity Disorder – x-linked recessive
- neurological disorder
    - Tuberous sclerosis – autosomal dominant
    - Neurofibromatosis – autosomal dominant
    - Rett syndrome – x-linked dominant
    - Tay Sachs disease – autosomal recessive
    - Gaucher disease – autosomal recessive
    - Familial dysautonomia – autosomal recessive
- cancer at a young age (<40)
    - Peutz-Jeghers syndrome (gastrointestinal) – autosomal dominant
    - Basal Cell Nevus syndrome (basal cell carcinomas) – autosomal dominant
    - Juvenile Polyposis syndrome (gastrointestinal) – autosomal dominant
- stillbirth/ neonatal death
    - Spinal muscular atrophy – autosomal recessive

- individuals that died in infancy/childhood
  - Tay Sachs disease – autosomal recessive
  - Canavan disease – autosomal recessive
  - Spinal muscular atrophy – autosomal recessive
  - Zellweger syndrome spectrum – autosomal recessive
  - Myotonic dystrophy – autosomal dominant
- joint laxity/recurrent dislocations
  - Marfan syndrome – autosomal dominant
  - Fragile X syndrome – x-linked (triplet repeat disorder)
  - Williams syndrome – autosomal dominant
  - Ehlers-Danlos syndrome – autosominal dominant
  - Osteogenesis Imperfecta – autosomal dominant
- bleeding/clotting disorders
  - Noonan syndrome – autosomal dominant
  - Factor V Leiden thrombophilia
- genetic anemia/thalassemia
  - Gaucher disease – autosomal recessive
- unusual skin growths/conditions
  - Tuberous sclerosis – autosomal dominant
  - Ehlers-Danlos syndrome – autosominal dominant
  - Fabry disease – x-linked
- multiple or unusual birthmarks
  - Neurofibromatosis type 1 – autosomal dominant
- epilepsy/seizures
  - Angelman syndrome – depends on mechanism, usually sporadic
  - Tay Sachs disease – autosomal recessive
  - Zellweger syndrome spectrum – autosomal recessive
  - mitochondrial disorders – various modes of inheritance
  - Rett syndrome – x-linked dominant
- energy problems/crises
  - mitochondrial disorders – various modes of inheritance
- thyroid problems
- unusual eye findings (tumors, lens dislocations/opacities, etc.)
  - Marfan syndrome – autosomal dominant
  - Neurofibromatosis – autosomal dominant
  - Tay Sachs disease – autosomal recessive
  - Osteogenesis Imperfecta – autosomal dominant
  - Fabry disease – x-linked
- problems with muscle tone or muscle spasticity
  - Angelman syndrome – depends on mechanism, usually sporadic
  - Tay Sachs disease – autosomal recessive
  - Canavan disease – autosomal recessive

- dental problems (absent/abnormally-shaped teeth)
  - Osteogenesis Imperfecta – autosomal dominant
  - Ectodermal dysplasia – various forms and modes of inheritance
- respiratory disease/chronic lung condition
  - Gaucher disease – autosomal recessive
  - Spinal muscular atrophy – autosomal recessive
  - Myotonic dystrophy – autosomal dominant
- overgrowth disorder
  - Beckwith-Wiedemann syndrome – sporadic, autosomal dominant
  - Sotos syndrome
- spinal problems
  - Marfan syndrome – autosomal dominant
  - Neurofibromatosis – autosomal dominant
  - Spinal muscular atrophy – autosomal recessive
  - Sotos syndrome – autosomal dominant
- abnormally developed hands/feet (ex: club foot)
  - Smith-Lemli-Opitz syndrome – autosomal recessive
  - Cornelia de Lange syndrome – autosomal dominant/x-linked
- patches of different colored hair/skin
  - Angelman syndrome – depends on mechanism, usually sporadic
  - Ectodermal dysplasia – various forms and modes of inheritance
  - Peutz-Jeghers syndrome (gastrointestinal) – autosomal dominant
- iron overload syndrome (hemochromatosis)
- kidney problems (extra/missing/polycystic)
  - Tuberous sclerosis – autosomal dominant
  - Beckwith-Wiedemann syndrome – sporadic, autosomal dominant
- multiple fractures/broken bones
  - Osteogenesis Imperfecta – autosomal dominant
- unusually tall for family
  - Marfan syndrome – autosomal dominant
  - Sotos syndrome – autosomal dominant
- unusually short for family
  - Noonan syndrome – autosomal dominant
  - Osteogenesis Imperfecta – autosomal dominant

Provider Family-History Follow-up Guide – by Condition

This guide is not intended to be diagnostic, but provides some additional information about possible genetic syndromes. For any given genetic syndrome, there are many features and this list only covers some of the more common symptoms/syndromes that might be picked up by family history. Additional information about genetic conditions is available at www.genetests.org. For additional information or if you have concerns about a patient's family history, please consult with a genetics professional.

- 22q11 deletion syndrome (VCFS/DiGeorge) (AD)
  - congenital heart defects, cleft palate, mental illness,
- Androgen Insensitivity Disorder (x-linked)
  - absent/missing puberty
- Angelman syndrome
  - autism, developmental delay, seizures, hypopigmentation
- Bardet-Biedl syndrome (AR)
  - vision loss, obesity, polydactyly, renal dysfunction, genital abnormalities
- Basal Cell Nevus syndrome (AD)
  - basal cell carcinomas, skeletal abnormalities (bifid ribs)
- Beckwith Wiedemann syndrome (AD/sporadic)
  - overgrowth (macrosomia), embryonal tumors, renal abnormalities, macroglossia
- Canavan disease (AR)
  - developmental delay, hypotonia, muscle spasticity, progressive leading to death in childhood
- Cornelia de Lange syndrome (AD/ x-linked)
  - hirsutism, autism, developmental delay, heart defect, upper limb abnormalies, hearing loss, growth retardation
- Ectodermal dysplasia (various forms and modes of inheritance)
  - can have any combination of the following: hearing loss, abnormal sweat, abnormal teeth, abnormal nails, sparse hair, pigmentary abnormalities
- Ehlers-Danlos syndrome (AD) – several types
  - joint laxity, skin fragility, abnormal scars, hyperelastic velvety skin, hypermobility/hyperextensibility, skeletal abnormalities
- Fabry disease (x-linked)
  - vascular cutaneous lesions, extremity pain, corneal opacities, proteinuria
- Fragile X syndrome (X-linked triplet repeat disorder)
  - developmental delay, joint laxity, macroorchidism, autistic features
- Gaucher disease (AR)
  - progressive neurologic disorder, bone disease, hepatosplenomegaly, anemia
- Hemochromatosis (AR)
  - iron overload syndrome
- Juvenile Polyposis syndrome (AD)
  - gastrointestinal cancer

- Marfan syndrome (AD)
    - tall stature, joint laxity, lens dislocations, scoliosis, aortic root dilation, mitral valve prolapse, pectus excavatum/carinatum
- mitochondrial disorders
    - hearing loss, vision loss, diabetes, seizures, energy crises,
- Myotonic dystrophy (AD)
    - myotonia, cataracts, muscle weakness, hypotonia
- Neurofibromatosis type 1 (AD)
    - skin findings (café au lait spots, neurofibromas, inguinal/axillary freckling), neurologic involvement, scoliosis, lisch nodules
- Neurofibromatosis type 2 (AD)
    - bilateral vestibular schwannomas, hearing loss, meningiomas
- Noonan syndrome (AD)
    - developmental delay, short stature, congenital heart defect, bleeding disorder
- Osteogenesis Imperfecta (AD)
    - joint laxity, brittle bones, dentinogenesis imperfecta, blue sclerae, skeletal abnormalities, short stature
- Peutz-Jeghers syndrome (AD)
    - gastrointestinal cancer, hyperpigmented macules
- Prader-Willi syndrome
    - developmental delay, failure to thrive as infant, excessive eating, obesity, short stature, behavior problems
- Retinitis pigmentosa
    - vision loss
- Rett syndrome (x-linked dominant)
    - autism, seizures, gait abnormalities
- Smith-Lemli-Opitz syndrome (AR)
    - developmental delay, cleft palate, hypospadias, polydactyly/syndactyly
- Sotos syndrome (AD)
    - learning problems, heart defect, overgrowth, scoliosis, seizures, renal abnormalities, behavior problems
- Spinal muscular atrophy
    - progressive muscle weakness, joint contractures, poor weight gain, scoliosis, infantile/childhood death, respiratory insufficiency
- Tay Sachs disease (AR)
    - progressive neurologic disorder, seizures, vision loss, muscle spasticity, leading to death in childhood/early adulthood
- Thrombophilias
    - clotting disorder
- Tuberous sclerosis (AD)
    - developmental delay, neurologic involvement (cortical tubers), cardiac rhabdomyomas,

- Usher syndrome (AR)
    - sensorineural hearing loss, vision loss, vestibular areflexia
- Williams syndrome (AD)
    - developmental delay, heart defect, joint laxity, wide full lips, cardiovascular disease, connective tissue disorder, hypotonia, failure to thrive, distinct personality
- Wolf-Hirschhorn syndrome
    - developmental delay, growth retardation, hypotonia, skeletal abnormalities, seizures, “Greek warrior helmet” facial appearance
- Zellweger syndrome spectrum (AR)
    - developmental delay, seizures, childhood death, liver cysts