Accuracy of physical examination of cardiovascular system in the diagnosis of common congenital heart diseases in children

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ABSTRACT

Background It is widely perceived that the value of physical examination in paediatric cardiology has diminished with the increasing availability of echocardiography. The accuracy of physical examination of cardiovascular system in children has not been systematically tested.

Methods This is a cross-sectional, diagnostic accuracy study from the paediatric cardiology clinic of a tertiary referral hospital in South India. A total of 545 children with 5 common cardiac conditions were included—normal, atrial septal defect, patent ductus arteriosus, ventricular septal defect (VSD) and VSD with pulmonic stenosis. Physical examination was documented by a paediatric cardiology fellow and a consultant who were blinded to previous investigations and to each other. The accuracy of physical examination of the fellow and the consultant was determined for each patient group by comparing with echocardiography. Interobserver agreement was calculated using kappa statistics.

Results Physical examination differentiated normal hearts from abnormal with an accuracy of 95.0% for fellows and 96.3% for consultants. For all abnormal hearts, the results for fellows and consultants, respectively, were as follows: sensitivity: 94.3%, 94.9%; specificity: 96.2%, 98.6%; accuracy: 95.0%, 96.3%; positive likelihood ratio: 24.8, 66.4 and negative likelihood ratio: 0.06, 0.05. There was good agreement between fellows and consultant for all patient groups (kappa: 0.72–1), except for large VSD (kappa: 0.232). Younger age and haemodynamically insignificant lesions were associated with incorrect diagnosis.

Conclusion This study underscores the utility of clinical examination in initial screening for commonly encountered congenital cardiac conditions even in the current era of echocardiography.

INTRODUCTION

History and physical examination are traditional tools that contribute substantially to arriving at an accurate diagnosis and strengthens our rapport with the patient. However, the importance of physical examination in patients with heart disease appears to be diminishing in recent times. Physical examination of cardiovascular system in children is challenged by faster heart rates, conducted airway sounds and limited patient cooperation. This has led to excessive use of more expensive investigations like echocardiography, which adds to healthcare costs and burdens health systems. In busy clinical settings, the overuse of echocardiography for relatively trivial situations can dilute, distract and limit the time available for comprehensive and nuanced assessments of significant structural lesions. Additionally, needless parental anxiety can perhaps be avoided by improved screening through a careful physical examination.

There is paucity of large studies demonstrating accuracy of physical examination in diagnosis of common cardiac diseases in adults and children. The reported accuracy of physical examination in published studies is quite variable. Clinical
examination, when treated like any other diagnostic
test, has two important characteristics—precision and
accuracy. Precision is determined by interobserver and
intraobserver agreement. Cohen’s kappa analysis is
widely used in assessing inter-rater agreement.16 17 Ac-
curacy is ascertained from sensitivity, specificity, positive
predictive value (PPV), negative predictive value (NPV),
positive likelihood ratio (LR+) and negative LR (LR−).18
Thresholds of >5 for LR+ and <0.3 for LR− have been
previously suggested for evaluation of utility of physical
signs.19

The primary objective of the study was to report the
diagnostic accuracy of physical examination of cardio-
vascular system by both a paediatric cardiology fellow as
well as a consultant in children with selected congenital
heart diseases using comprehensive echocardiography
as a gold standard. The secondary objective of the study
was to assess interobserver agreement between a fellow
and consultant for diagnosis of selected congenital heart
diseases by physical examination.

METHODS
This is a diagnostic accuracy study conducted in the
setting of paediatric cardiology outpatient clinic of a
tertiary referral centre in South India from May 2020 to
October 2021. Patients or the public were not involved
in the design, or conduct, or reporting, or dissemination
plans of the research.

Selection and description of participants
Patients under the age of 18 years with a diagnosis of
‘normal heart’, ‘atrial septal defect’ (ASD), ‘patent ductus
arteriosus’ (PDA), ‘ventricular septal defect’ (VSD) and
‘VSD with pulmonic stenosis’ (VSD-PS) visiting the paediat-
ric cardiology clinic for the first time were included. We
cchose to evaluate only the above lesions because they are
common and have relatively specific physical signs.

The sample size was calculated based on the results
of the pilot study, conducted on 20 patients. The sensi-
tivity of the physical examination in diagnosis of cardiac
diseases was 77.9% using echocardiography as the gold
standard. Based on the sensitivity with 10% relative preci-
sion and 95% CI, the minimum number of positive cases
(children with listed CHD) was calculated to be 66. The
total number required for the study was calculated to be
147 subjects based on the proportion of children with
disease (45%) in the pilot study. We recruited a larger
number of subjects (545) to allow for subgroup analysis
in each category.

Technical information
Six paediatric cardiology fellows and four paediatric card-
iology consultants took part in the study. All paediatric
cardiology fellows had a minimum of 6 months exposure
to paediatric cardiac patients during their training years
after completing a 3-year residency in general paediat-
rics. The patients were first examined by a paediatric
cardiology fellow and then by a paediatric cardiology
consultant. Both observers used a non-digital (Littmann
Master Cardiology IV) stethoscope. The detailed findings
of inspection, palpation and auscultation of all patients
were documented in a physical examination form (see
online supplemental material). Both observers were
allowed enough time to do a full physical examination
in a quiet setting and were blinded to history and the
prior investigations, each other’s physical examination
findings and to the echocardiography findings. Their
findings and diagnosis was documented in a structured
physical examination form that was filed immediately
thereafter. A comprehensive echocardiogram was then
performed by a consultant or paediatric cardiology fellow
(under supervision) using a high-end echocardiography
machine (Philips EPIQ /iE33).

We have a pool of four consultants and six fellows. For
this study we did not mandate that a separate group of
consultants or fellows do the echocardiograms. There-
dependentcl, there were instances when the person conducting
the physical examination also did the echocardiograms.
We recognise that this may bring in a bias. However, we do
have a standardised protocol for performing, recording
and interpreting echocardiograms. Oral sedation with
weight appropriate dose of tricloflos syrup (500 mg/5
mL) was given to uncooperative children.

Statistics
Statistical analyses were conducted using SPSS V.20.0
for Windows (IBM). Categorical data were described as
numbers and percentage. Age had skewed distribution
which was described as median with an IQR. Echocar-
diography was considered the gold-standard diagnostic
test. We report the sensitivity, specificity, PPV, NPV, accu-
racR, LR+ and LR− of clinical examination in compar-
ison to the echocardiography (gold standard). An inter-
rater reliability analysis using the kappa statistics was
performed to determine concurrence among observers.
CIs for sensitivity, specificity and accuracy were calculated
using Clopper-Pearson CIs. CIs for the LR were calcu-
lated using the log method and for the predictive values
using the standard logit CIs.20 21 CIs for kappa values were
calculated using generic formula (estimate±1.96 SE) for
95% CIs.

RESULTS
Complete evaluation by one consultant and one fellow
was possible in 935 patients of which we excluded 390
because they had lesions other than those chosen for
the study. Hence, 545 newly registered patients with the
aforementioned diagnoses were included in the study.

Demography
A total of 297 (54.5%) children were females and 191
(35.1%) were infants. There were 210 children (38.5%)
with normal heart. The median age of the patients was
2 years (IQR 7.2 months to 6.0 years, range 7 days to

18.0 years). ASD (n=130, 23.9%) constituted the biggest group among the children with abnormal heart, followed by those with PDA (n=90, 16.5%). Age and sex distribution of the patients stratified by their cardiac diagnosis is shown in Table 1.

### Accuracy of physical examination by fellow
The accuracy of physical examination by the pediatric cardiology fellow for differentiating normal from abnormal heart was 95.0% with a sensitivity of 96.2% and a specificity of 94.3%. PPV of physical examination for normal heart was 91.4% and NPV was 97.5%. LR+ for diagnosis of normal heart was 17.0 and LR− was 0.04. Eight of 210 children (3.8%) with normal heart were mislabelled as having heart disease on physical examination and 19 children with heart disease were incorrectly diagnosed as normal heart by physical examination.

There were 335 children (61.5%) with abnormal heart; 33 children (9.9%) with abnormal heart were missed on physical examination which included ASD 10, PDA 13, VSD 5 and VSD-PS 5. Sixteen children were incorrectly diagnosed clinically as ASD 2, PDA 2 and VSD 12. The LR+ for diagnosis of abnormal heart by fellow was 24.8 and LR− was 0.06. ASD was the most common diagnosis among the abnormal hearts. For ASD, physical examination by pediatric cardiology fellow had accuracy 97.8% with sensitivity 92.3% and specificity of 99.5%. LR+ for diagnosis of ASD was 191.5 and LR− was 0.08. VSD-PS group had the best accuracy of 99.1% with sensitivity 86.1%, specificity 100%, PPV 100% and NPV 99.1%. There were 191 infants in the study. The accuracy of physical examination by fellow for differentiating normal from abnormal heart in infants was 93.2% with a sensitivity of 91.9% and a specificity of 93.8%. Table 2 shows the accuracy and LR of physical examination by fellow for various conditions studied.

### Accuracy of physical examination by consultant
The study showed accuracy of physical examination by pediatric cardiology consultant for differentiating normal from abnormal heart to be 93.2% with a sensitivity of 92.3% and a specificity of 93.8%. Table 2 shows the accuracy and LR of physical examination by consultant for various conditions studied.
The median age of incorrectly diagnosed patients by the fellows as well as consultants was significantly less than the correctly diagnosed patient group (9.6 months vs 2 years, p=0.04 for residents and p=0.015 for consultants). Table 5 shows the details of all missed diagnoses by fellow and consultant. Severe heart disease was nondiagnosed in 12.2% by the consultants and 13.6% by the fellows. Seventeen out of 32 (53.1%) patients misdiagnosed by the fellows had a normal heart, whereas 20 out of 32 (62.5%) patients misdiagnosed by the consultants had a normal heart. The accuracy for the diagnosis of heart disease was 95.8% with a sensitivity of 98.4% and a specificity of 94.6%. Table 3 shows the accuracy and LR of physical examination for various cardiac conditions.

### Table 3: Accuracy of physical examination by consultant versus echocardiography

<table>
<thead>
<tr>
<th>Condition</th>
<th>Sensitivity in % (95% CI)</th>
<th>Specificity in % (95% CI)</th>
<th>PPV in % (95% CI)</th>
<th>NPV in % (95% CI)</th>
<th>Accuracy in % (95% CI)</th>
<th>LR+ (95% CI)</th>
<th>LR− (95% CI)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal heart</td>
<td>98.6 (95.9 to 99.7)</td>
<td>94.9 (92.0 to 97.0)</td>
<td>92.4 (88.5 to 95.1)</td>
<td>99.1 (97.2 to 95.1)</td>
<td>96.3 (94.4 to 97.7)</td>
<td>19.4 (12.2 to 30.9)</td>
<td>0.02 (0.00 to 0.05)</td>
</tr>
<tr>
<td>Abnormal heart*</td>
<td>94.9 (92.0 to 97.0)</td>
<td>98.6 (95.9 to 99.7)</td>
<td>99.1 (97.2 to 95.1)</td>
<td>92.4 (88.5 to 95.1)</td>
<td>96.3 (94.4 to 97.7)</td>
<td>66.4 (21.6 to 204.4)</td>
<td>0.05 (0.03 to 0.08)</td>
</tr>
<tr>
<td>ASD</td>
<td>95.4 (90.2 to 98.3)</td>
<td>99.5 (98.3 to 99.9)</td>
<td>98.4 (93.7 to 99.6)</td>
<td>98.6 (96.9 to 99.3)</td>
<td>98.5 (97.1 to 99.4)</td>
<td>197.9 (49.6 to 789.2)</td>
<td>0.05 (0.02 to 0.10)</td>
</tr>
<tr>
<td>PDA</td>
<td>87.8 (79.2 to 93.7)</td>
<td>100.0 (99.2 to 100.0)</td>
<td>97.6 (95.9 to 98.6)</td>
<td>98.0 (96.4 to 98.9)</td>
<td>197.6 (49.6 to 789.2)</td>
<td>0.12 (0.07 to 0.21)</td>
<td>† (0.07 to 0.10)</td>
</tr>
<tr>
<td>VSD</td>
<td>92.4 (84.2 to 97.2)</td>
<td>98.5 (96.9 to 99.4)</td>
<td>91.2 (83.3 to 95.6)</td>
<td>98.7 (97.3 to 99.4)</td>
<td>97.6 (95.9 to 98.7)</td>
<td>61.5 (29.4 to 128.7)</td>
<td>0.08 (0.04 to 0.17)</td>
</tr>
<tr>
<td>VSD-PS</td>
<td>91.7 (77.5 to 98.3)</td>
<td>100.0 (99.3 to 100.0)</td>
<td>99.4 (98.3 to 99.8)</td>
<td>99.4 (98.4 to 99.9)</td>
<td>99.4 (98.4 to 99.9)</td>
<td>0.08 (0.03 to 0.25)</td>
<td>† (0.07 to 0.10)</td>
</tr>
<tr>
<td>Normal heart for age &lt;1 year</td>
<td>98.4 (91.3 to 100.0)</td>
<td>94.6 (91.1 to 97.8)</td>
<td>89.7 (80.9 to 94.7)</td>
<td>99.2 (94.6 to 99.9)</td>
<td>95.8 (91.9 to 98.2)</td>
<td>18.1 (8.8 to 37.3)</td>
<td>0.02 (0.0 to 0.12)</td>
</tr>
<tr>
<td>Abnormal heart for age &lt;1 year</td>
<td>94.6 (89.1 to 97.8)</td>
<td>98.4 (91.3 to 100.0)</td>
<td>99.2 (94.6 to 99.9)</td>
<td>89.7 (80.9 to 94.7)</td>
<td>95.8 (91.9 to 98.2)</td>
<td>58.6 (8.4 to 409.9)</td>
<td>0.06 (0.03 to 0.11)</td>
</tr>
</tbody>
</table>

*Abnormal heart includes ASD, PDA, VSD and VSD-PS.
†Approaches infinity as specificity is 100.
ASD, atrial septal defect; LR−, negative likelihood ratio; LR+, positive likelihood ratio; NPV, negative predictive value; PDA, patent ductus arteriosus; PPV, positive predictive value; VSD, ventricular septal defect; VSD-PS, VSD with pulmonic stenosis physiology.
DISCUSSION

Our results showed that the accuracy of physical examination, by the paediatric cardiology fellow and the consultant, for differentiating normal from abnormal heart exceeded 95%. There was good interobserver agreement between the fellow and consultant’s diagnosis for all patient groups except for large VSD.

The strength of our study is in the number of patients in the study and its focus on the paediatric population with common congenital heart diseases. In a study of 104 patients (age ranging from 2 to 85 years) by Patel et al., authors demonstrated ‘almost perfect’ agreement between auscultation of the heart by a senior cardiologist and echocardiography for the diagnosis of mitral stenosis and VSD and substantial agreement for pulmonary stenosis, aortic stenosis and ASD.14

The ability to correctly differentiate an innocent murmur from a pathological murmur on physical examination helps avoid unnecessary referrals for echocardiography, the resultant parental anxiety and healthcare costs.9–13 In our study, 96.2% of children with a normal

### Table 4
<table>
<thead>
<tr>
<th>Condition</th>
<th>Measure of agreement (kappa)</th>
<th>95% CI, lower limit-upper limit</th>
<th>P value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal heart</td>
<td>0.951</td>
<td>0.924 to 0.978</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>ASD</td>
<td>0.735</td>
<td>0.488 to 0.9982</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>PDA</td>
<td>0.904</td>
<td>0.773 to 1.35</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Large VSD</td>
<td>0.232</td>
<td>−0.225 to 0.689</td>
<td>0.1</td>
</tr>
<tr>
<td>Restrictive VSD</td>
<td>1.000</td>
<td>NA</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>VSD - PS physiology</td>
<td>0.721</td>
<td>0.360 to 1.082</td>
<td>&lt;0.001</td>
</tr>
</tbody>
</table>

Interpretation of kappa values—value ≤0 indicates no agreement, 0.01–0.20 indicates slight agreement, 0.21–0.40 fair, 0.41–0.60 as moderate, 0.61–0.80 as substantial and 0.81–1.00 indicates almost perfect agreement.16 17

ASD, atrial septal defect; NA, not applicable; PDA, patent ductus arteriosus; VSD, ventricular septal defect; VSD-PS, VSD with pulmonic stenosis physiology.

### Table 5
<table>
<thead>
<tr>
<th>Fellow</th>
<th>No of missed diagnoses</th>
<th>Median age in months (IQR)</th>
<th>Patient details</th>
<th>Consultant</th>
<th>No of missed diagnoses</th>
<th>Median age in months (IQR)</th>
<th>Patient details</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal heart</td>
<td>8</td>
<td>9.3 (6–54)</td>
<td>Systolic murmur in left upper sternal border</td>
<td>3</td>
<td>12 (6.08–14)</td>
<td>Systolic murmur in left upper sternal border</td>
<td></td>
</tr>
<tr>
<td>ASD</td>
<td>10</td>
<td>7.2 (2.4–9.6)</td>
<td>6 had small ASD (&lt;7 mm), 1 had cyanosis because of large ASD amounting to common atrium 3 were infants with large ASD diagnosed as VSD</td>
<td>6</td>
<td>0.6 (0.21–0.8)</td>
<td>5 had small ASD (&lt;7 mm), One was a 3-month-old baby with a large ASD</td>
<td></td>
</tr>
<tr>
<td>PDA</td>
<td>13</td>
<td>21.6 (8.4–60)</td>
<td>9 had tiny PDA A 6-month-old baby with 2.5 mm PDA which was clinically diagnosed as ‘normal’ 3 large PDA were clinically diagnosed as large VSD</td>
<td>11</td>
<td>1.8 (0.7–5)</td>
<td>9 had tiny PDA 2 large PDA were diagnosed as large VSD</td>
<td></td>
</tr>
<tr>
<td>VSD</td>
<td>5</td>
<td>4.8 (4.8–48)</td>
<td>2 small VSDs 1 large VSD was diagnosed as large PDA 1 large VSD was diagnosed as restrictive VSD One 22-day-old baby with large VSD was diagnosed as normal</td>
<td>6</td>
<td>4 (2.04–9)</td>
<td>2 small VSDs were diagnosed as normal. 2 large VSD were diagnosed as restrictive VSD. One 22-day-old baby with large VSD was diagnosed as normal. Another 2-month-old baby was diagnosed as pulmonic stenosis</td>
<td></td>
</tr>
<tr>
<td>VSD-PS</td>
<td>5</td>
<td>3 (0.24–18)</td>
<td>All were acyanotic and had a systolic murmur at left upper sternal border</td>
<td>3</td>
<td>0.25 (0.02–1.5)</td>
<td>All were acyanotic and had a systolic murmur at left upper sternal border</td>
<td></td>
</tr>
</tbody>
</table>

Total | 41 | 29 |

ASD, atrial septal defect; PDA, patent ductus arteriosus; VSD, ventricular septal defect; VSD-PS, VSD with pulmonic stenosis physiology.
heart and 94.3% of children with abnormal heart were correctly identified by the fellow. In the study of auscultation skills of all paediatric fellows by Mahnke et al, with focused cardiology training and computer based practice, the diagnostic accuracy of the innocent murmur increased from 35% to 65%. In our study, diagnostic accuracy of physical examination for normal heart was >95% for both fellows and consultants. The ability to diagnose normal from abnormal heart can thus be improved by training and with experience.4–7

There was excellent interobserver agreement between the fellow and consultant’s diagnosis for all patient groups except for large VSD which reinforces the reliability of physical examination as a diagnostic tool (table 4). This also shows that junior doctors can improve their clinical skills with practice so as to reach diagnostic accuracy of the consultants.

As expected, incorrectly diagnosed patients were significantly younger than the correctly diagnosed children (9.6 months vs 2 years, p<0.04). The examination is particularly difficult in infants and toddlers and could contribute to incorrect diagnosis.

Of the 41 diagnoses missed on physical examination by the fellow, 17 patients had conditions that are not haemodynamically significant and which can be hard to detect clinically. These include nine with tiny PDA, six small ASD and two small VSD. Sixteen (of 29) of the missed diagnosis by consultant were small ASD, tiny PDA or small VSD. The physical examination by fellow was, at times, challenging because of noisy surroundings of the busy outpatient clinic or if the child was crying. Consultants often had the advantage of quieter surroundings and sedated child as examination was performed just before echocardiography. The total number of clinically significant diagnoses missed by fellow was 24 out of 545 (4.4%). The total number of clinically significant diagnoses missed by the consultant was 13 out of 545 (2.4%). This result suggests that very few clinically significant cardiac conditions in children were likely to be missed by careful physical examination if performed by trained observers in an optimal setting.

The accuracy levels and interobserver agreement of physical examination in our study are encouraging and meet the criteria of a good diagnostic test.18 19 However, it is necessary to acknowledge the following limitations that may limit the generalisability of our results.

First, the fellows involved in the study were at various levels of their training in paediatric cardiology. All the fellows had 3–6 years of prior training in general paediatrics and 6 months to 2 years in paediatric cardiology. The high accuracy of the fellows might be due to their familiarity with the studied CHDs. Whether a general paediatrician or family doctor in the community can achieve similar results without focused cardiology training, remains to be shown.

Second, the study only looked at four common congenital heart diseases. The study’s results cannot be generalised to other congenital heart diseases or valvular heart diseases. Also, the numbers of patients in the large VSD and VSD-PS group were small and therefore the results in these groups are not as robust.

Third, the study was conducted in a paediatric cardiology outpatient clinic of a tertiary care hospital where the observers are more biased towards suspecting and detecting CHD. Such patients are also more likely to have heart disease. Hence, the pretest probability of heart disease is high in these patients. Therefore, these results cannot be generalised to children attending a general paediatrics clinic or community healthcare setting.

Finally, the results of this study cannot be extrapolated to inpatient settings, for example, neonates who are being evaluated in Neonatal Intensive Care Unit settings.

CONCLUSION

Our results demonstrate that physical examination remains a useful tool in the armamentarium of the cardiologist. Given the high accuracy of physical examination in differentiating normal heart from abnormal, it may be possible to limit the number of unnecessary echocardiograms in busy outpatient settings, thereby reducing healthcare expenditure. Further, careful physical examination can serve as a useful screening tool in paediatric office settings especially in situations where there are limitations in access to echocardiography. While additional studies are needed to test general paediatricians with limited training and assess the impact of targeted training in cardiac examination, our results suggest that there is value to investing in acquisition of skills in cardiovascular examination. The evidence generated by our study suggests that the obituary written for the stethoscope is perhaps not justified as yet.

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Patient consent for publication Not applicable.

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