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 heart, 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, inability to perform assessment in some cases, and makes echocardiography more preferred. This study underscores the utility of clinical examination in initial screening for commonly encountered congenital cardiac conditions even in the current era of echocardiography.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ The accuracy of physical examination of cardiovascular system in children has not been systematically tested using echocardiography as a gold standard.

WHAT THIS STUDY ADDS

⇒ Careful physical examination can distinguish normal heart from abnormal with a high degree of accuracy and can serve as a useful screening tool in paediatric office settings. Physical examination is also quite accurate in identifying common congenital heart diseases in children.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ Physical examination of the cardiovascular system is a potentially valuable diagnostic tool that must be systematically tested in diverse clinical settings. When applied correctly, it could serve to rationalise the use of echocardiography and potentially enable substantial reductions in healthcare costs.
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. Accur

Thresholds of >5 for LR+ and <0.3 for LR− have been previously suggested for evaluation of utility of physical signs. The primary objective of the study was to report the diagnostic accuracy of physical examination of cardiovascular 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 paediatric cardiology clinic for the first time were included. We chose 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 sensitivity 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 precision 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 cardiology 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 paediatrics. 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. Therefore, 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 triclofos 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. Echocardiography was considered the gold-standard diagnostic test. We report the sensitivity, specificity, PPV, NPV, accuracy, LR+ and LR− of clinical examination in comparison 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 calculated using the log method and for the predictive values using the standard logit CIs. 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
Alawani SS, et al. BMJ Paediatrics Open 2022;6:e001595. doi:10.1136/bmjpo-2022-001595

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 paediatric 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 paediatric 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 89.9% and specificity with 93.2. 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 paediatric cardiology consultant for differentiating

| Table 1 | Age and sex distribution of patients by individual diagnosis |
|---------|-------------------------------------------------------------|
|          | No of cases n (%) | Females n (%) | Age—median age in years (IQR) |
| Normal heart | 210 (38.5) | 101 (48.1) | 3 (0.75, 10.0) |
| ASD | 130 (23.9) | 76 (58.5) | 4 (3.0, 7.0) |
| PDA | 90 (16.5) | 65 (72.2) | 1 (0.5, 2.1) |
| Large VSD | 49 (8.9) | 28 (57.1) | 0.4 (0.2, 0.7) |
| Restrictive VSD | 30 (5.5) | 14 (46.7) | 2.5 (0.75, 7.0) |
| VSD+PS | 36 (6.6) | 13 (36.1) | 0.5 (0.2, 1.5) |
| Total | 545 | 297 (54.5) | 2 (0.02–18.0) |

ASD, atrial septal defect; PDA, patent ductus arteriosus; VSD, ventricular septal defect; VSD+PS, VSD with pulmonic stenosis.
normal from abnormal heart was 96.3% with sensitivity 98.6%, specificity 94.9%, PPV 92.4% and NPV 99.1%. The LR+ for diagnosis of normal heart was 19.4 and LR− was 0.02. Three children with normal heart were mislabelled as having heart disease by physical examination. Seventeen children with heart disease were incorrectly diagnosed as normal heart by physical examination.

For ASD, physical examination by paediatric cardiology consultant had an accuracy of 98.5% with sensitivity 95.4%, specificity 99.5%, PPV 98.4% and NPV 99.3%. VSD-PS group had the best accuracy of 99.4% with sensitivity of 91.7%, specificity of 100%, PPV 100% and NPV 99.4%.

The accuracy of physical examination by consultant for differentiating normal from abnormal heart for infants (n=191) 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 by consultant for various conditions studied in comparison with gold standard test of echocardiography.

Interobserver agreement between fellows and consultant
Kappa analysis showed perfect agreement between fellow in training and consultant for restrictive VSD (kappa=1) and near-perfect agreement for diagnoses of normal heart (kappa=0.951) and PDA (kappa=0.904). There was substantial agreement between the two observers for the diagnoses of ASD (kappa=0.735) and VSD-PS physiology (kappa=0.721). There was only fair agreement between the two observers for diagnosis of large VSD (kappa=0.232). Table 4 shows the kappa values for each cardiac condition.

Analysis of missed diagnoses
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 9.6 months vs 2 years, p=0.016 for consultants). Table 5 shows the details of all missed diagnoses by fellow as well as consultant. Seventeen (of 41; 41%) patients missed by fellow on physical examination had conditions which were clinically insignificant. These include nine with tiny PDA, six small ASD and two small VSDs. Sixteen of 29 (55%) of the diagnoses missed by consultant were small ASD, tiny PDA and small VSD. Twenty-four significant conditions were incorrectly diagnosed by the fellow that included normal heart eight, common atrium one, large ASD three, large PDA four, large VSD three and VSD-PS physiology five. Thirteen significant conditions that were incorrectly diagnosed by consultant were normal heart three, large ASD one, large PDA two, large VSD four and VSD-PS physiology three.

| Condition            | Accuracy in % (95% CI) | Sensitivity in % (95% CI) | Specificity in % (95% CI) | PPV in % (95% CI) | NPV in % (95% CI) | LR+ (95% CI) | LR− (95% CI) |
|----------------------|------------------------|---------------------------|---------------------------|------------------|------------------|---------------|---------------|
| Normal heart         | 96.8 (95.9 to 97.7)    | 94.9 (92.0 to 97.0)       | 96.6 (95.9 to 97.7)       | 99.1 (97.2 to 99.7) | 96.3 (94.4 to 97.7) | 19.4 (12.2 to 30.9) | 0.02 (0.00 to 0.05) |
| Abnormal heart*      | 94.9 (92.0 to 97.0)    | 96.6 (95.9 to 97.7)       | 99.1 (97.2 to 99.7)       | 96.3 (94.4 to 97.7) | 98.6 (97.1 to 99.4) | 197.9 (49.6 to 789.2) | 0.12 (0.07 to 0.21) |
| ASD                  | 96.4 (90.2 to 98.3)    | 98.6 (95.9 to 99.7)       | 99.4 (97.1 to 99.8)       | 98.6 (97.1 to 99.4) | 98.6 (97.1 to 99.4) | 61.5 (23.4 to 128.7) | 0.08 (0.04 to 0.25) |
| VSD                  | 91.8 (79.3 to 94.3)    | 98.5 (96.9 to 99.4)       | 98.7 (95.9 to 99.8)       | 98.7 (95.9 to 99.8) | 98.7 (95.9 to 99.8) | 18.1 (6.8 to 31.3) | 0.02 (0.03 to 0.12) |
| VSD-PS               | 91.7 (77.5 to 98.2)    | 98.6 (95.9 to 99.7)       | 98.7 (95.9 to 99.7)       | 98.7 (95.9 to 99.7) | 97.6 (96.5 to 98.7) | 58.6 (18.4 to 409.9) | 0.06 (0.03 to 0.11) |

*Abnormal heart includes ASD, PDA, VSD and VSD-PS.
†Abnormal heart includes ASD, PDA, VSD and VSD-PS.
‡Abnormal heart includes ASD, PDA, VSD and VSD-PS.
§Abnormal heart includes ASD, PDA, VSD and VSD-PS.
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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.

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. In our study, 96.2% of children with a normal heart had a systolic murmur in left upper sternal border.

| Condition      | Measure of agreement (kappa) | 95% CI, lower limit-upper limit | P value |
|----------------|-----------------------------|--------------------------------|---------|
| Normal heart   | 0.951                       | 0.924 to 0.978                 | <0.001  |
| ASD            | 0.735                       | 0.488 to 0.9982                | <0.001  |
| PDA            | 0.904                       | 0.773 to 1.35                  | <0.001  |
| Large VSD      | 0.232                       | −0.225 to 0.689                | 0.1     |
| Restrictive VSD| 1.000                       | NA                             | <0.001  |
| VSD - PS physiology | 0.721 | 0.360 to 1.082                 | <0.001  |

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 Details of missed diagnoses |
|------------------------------------|
| **Fellow**                      | **Consultant**                      |
|                                   | No of missed diagnoses | Median age in months (IQR) | Patient details | No of missed diagnoses | Median age in months (IQR) | Patient details |
| Normal heart                     | 8                             | 9.3 (6 – 54)               | Systolic murmur in left upper sternal border | 3                             | 12 (6.08–14) | Systolic murmur in left upper sternal border |
| ASD                              | 10                            | 7.2 (2.4–9.6)              | 6 had small ASD (<7 mm), 1 had cyanosis because of large ASD amounting to common atrium, 3 were infants with large ASD diagnosed as VSD | 6                             | 0.6 (0.21–0.8) | 5 had small ASD (<7 mm), One was a 3-month-old baby with a large ASD |
| PDA                              | 13                            | 21.6 (8.4–60)              | 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 | 11                            | 1.8 (0.7–5)  | 9 had tiny PDA, 2 large PDA were diagnosed as large VSD |
| VSD                              | 5                             | 4.8 (4.8–48)               | 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 | 6                             | 4 (2.04–9)  | 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 |
| VSD- PS                          | 5                             | 3 (0.24–18)                | All were acyanotic and had a systolic murmur at left upper sternal border | 3                             | 0.25 (0.02–1.5) | All were acyanotic and had a systolic murmur at left upper sternal border |
| **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 33% 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.

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 perhaps 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 VSDs. 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. 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 valvar 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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Acknowledgements Authors are sincerely grateful to Dr Balaji Seshadri, Dr Balu Vaidyanathan and Dr Mahesh Kappanayil for critically revising the manuscript. Special thanks to Renjitha Bhaskaran for helping in statistical analysis.
Contributors SSA, MR and RKK contributed to design of the study, acquisition of data, drafting and revising the manuscript. AS analysed and interpreted the data and revised the manuscript. RKK accepts full responsibility for the work and/or the conduct of the study, had access to the data, and controlled the decision to publish. All authors approved the final manuscript.
Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.
Competing interests No, there are no competing interests.
Patient and public involvement Patients and/or the public were not involved in the design, or conduct, or reporting, or dissemination plans of this research.
Patient consent for publication Not applicable.
Ethics approval This study involves human participants and was approved by Institutional Review Board, Amrita Institute of Medical Sciences. Reference number- IRB-AMS-2020-237. Participants gave informed consent to participate in the study before taking part.
Provenance and peer review Not commissioned; externally peer reviewed.
Data availability statement Data are available on reasonable request.
Supplemental material This content has been supplied by the author(s). It has not been vetted by BMJ Publishing Group Limited (BMJ) and may not have been
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